

DEPARTMENT OF HEALTH AND HUMAN SERVICES

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

Third Meeting

Monday,
March 1, 2004

Congressional Ballroom I-III
Bethesda Marriott
5151 Pooks Hill Road
Bethesda, Maryland

IN ATTENDANCE:

Chair

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C O N T E N T S

Welcome and Opening Remarks

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

Review of Process and Outcome of Inter-Meeting
Priority Setting Project and Presentation of
12 High Priority Issues

Emily Winn-Deen, Ph.D.
Chair, Inter-Meeting Task Force

Discussion of Review of Process and Outcome
of Inter-Meeting Priority Setting Project

Discussion and Votes on Priority Issues

Access

Coverage and Reimbursement

Education and Workforce

Large Population Studies

Public Awareness

Public Comment

Margaret Gulley, M.D.
College of American Pathologists

Dr. Judith Lewis
International Society of Nurses in Genetics

Sharon Terry, M.A.
Genetic Alliance

Andrea Ferreira-Gonzalez, Ph.D.
Association for Molecular Pathology

C O N T E N T S

Session on Coverage and Reimbursement for
Genetic Technologies and Services

Private Health Insurance Coverage and Payment
Policies and Decisionmaking Process for
Genetic Technologies and Services

Michele Schoonmaker, Ph.D.
Specialist in Genetics
Congressional Research Service

Genetic Services: The HMO Model

Ronald Bachman, M.D.
Chief, Genetics Department
Kaiser Permanente of Northern California

Providers' Perspectives on Reimbursement of
Genetic Technologies and Services

A Laboratorian's Perspective

Andrea Ferreira-Gonzalez, Ph.D.
Associate Professor and Director,
Molecular Diagnostics Laboratory
Virginia Commonwealth University

A Clinician's Perspective

Marc Williams, M.D.
Pediatrician and Medical Geneticist
Gundersen Lutheran Medical Center

C O N T E N T S

Medicare Coverage and Payment Policies and
Decisionmaking Processes for Genetic
Technologies and Services

Medicare Coverage Policies and Decisions

Sean Tunis, M.D., M.Sc.
Chief Medical Officer
Centers for Medicare and Medicaid Services

Medicare Payment Rates and Decisions

Donald Thompson, M.S.
Director of Ambulatory Services
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Cost-Effectiveness Determinants and Data Needs

David Veenstra, Pharm.D., Ph.D.
Assistant Professor
Department of Clinical Pharmacy
University of California-San Francisco

Roundtable Discussion

P R O C E E D I N G S

(8:40 a.m.)

1 DR. McCABE: Good morning, everyone, and welcome to the third meeting
2 of the Secretary's Advisory Committee on Genetics, Health, and Society.

3 The public was made aware of this meeting through notices in the Federal
4 Register, as well as announcements on the SACGHS website and listserv.

5 First I'd like to take note of two new ex officio appointments to the
6 committee. We're extremely pleased to welcome Mr. Matthew Daynard, senior attorney for the
7 Federal Trade Commission. Mr. Daynard briefed us in October about the FTC's mission; and
8 also Dr. Ellen Fox, director of the National Center for Ethics at the Department of Veterans
9 Affairs. Thank you both for participating.

10 We're extremely pleased that your agencies regard the work of SACGHS as
11 important and relevant to their missions. As with all of our ex officio members, we very much
12 appreciate the time and effort that you make to participate in the work of the committee and the
13 expertise and perspective that you all bring to our deliberations.

14 Before I review today's agenda, I'd like to remind everyone that at the end of
15 the October meeting, the committee decided that it was important to engage in a systematic
16 process to determine the priority issues that should be the focus of our work during the next
17 year. An inter-meeting task force was formed to guide the issue identification process and help
18 plan this meeting.

19 During the past four months, the task force has been hard at work on this
20 important project. I'd like to take this opportunity to thank the members of the task force for
21 your efforts on the committee's behalf. Emily Winn-Deen, who chaired the group; Cindy
22 Berry; Barbara Harrison; Debra Leonard; Reed Tuckson; and Hunt Willard, all members of the
23 task force.

24 In a moment we will hear more about the activities of the task force from
25 Emily Winn-Deen.

26 I also want to say a word about the 12 issue briefs in Tab 4 of your briefing
27 books. They were prepared by SACGHS staff under the direction of the task force. The briefs
28 have provided us with precise background information about the issues, given us deeper
29 understanding of their policy implications, and helped prepare us for our deliberations. I'd like
30 to thank the SACGHS staff for your hard work and dedication in preparing the briefs. Our
31 thanks and compliments especially to the principal authors, Amanda Sarata, Fay Shamanski,
32 Suzanne Goodwin, and Krista Crider. Of course, since Sarah prepared these remarks for me,
33 she did not give herself credit for having supervised all of that work, so thank you because we
34 know how much, Sarah, you put into this as well.

35 Actually, Hunt Willard and I were commenting that before the session this
36 morning, that we think these are such excellent documents at framing these issues that we
37 really think that there ought to be an exploration of publication in some forum to get this work
38 out so even more of the public could see this.

39 The primary goals of this meeting are, one to identify the top one to three
40 issues that will be our focus for the coming year; two, to develop a work plan, and three, to
41 begin deliberations on the first issue. The first half of today's agenda will be devoted to hearing
42 about the work of the task force and advancing our priority-setting process. In order to
43 organize the forthcoming discussion and engage the committee's current views on the
44 prioritization of issues, we will take a straw vote before today's first break. Then, with the
45

1 benefit of today's discussion, we will take a second straw vote at the end of the day. You'll find
2 in your table folders two ballots to use for these straw votes. These are in the white briefing
3 folders, this one, so that you can find them and be prepared for those straw votes.

4 In the afternoon we will hear a series of presentations and will hold a
5 roundtable discussion on coverage and reimbursement of genetic technologies and services.
6 Questions about coverage and reimbursement were raised in the October meeting, and a
7 number of members suggested that an in-depth briefing might be in order. In addition,
8 coverage and reimbursement is one of the 12 priority issues under consideration, and during the
9 inter-meeting process it was ranked among the top three issues.

10 This afternoon's session will explore public and private health insurance
11 coverage and payment policies for genetic technologies, the perspectives of service providers
12 and the financing and economic considerations with respect to these technologies.

13 Tomorrow, after we make our final determinations about the top three
14 priority issues, we will develop a long-range work plan for accomplishing the three projects.
15 Then we will begin deliberations on the top priority issues. We hope to make enough progress
16 on the first issue to be able to outline preliminary recommendations to the Secretary.

17 Let me also point out that we have public comment sessions each day. We
18 will hear testimony immediately following lunch today and first thing tomorrow morning. We
19 have a number of individuals, five, who have registered to address us. If there are others here
20 who wish to provide comments, please inform the individuals at the registration desk so that
21 you can get onto the list.

22 I'll now turn to Sarah Carr for an important reminder about the conflict of
23 interest rules.

24 Sarah?

25 MR. CARR: Thank you, Ed.

26 Being a member of this committee makes you a special government
27 employee -- I'm speaking to the members of the committee now -- and thereby subject to rules
28 of conduct that apply to government employees. The rules and regulations are explained in a
29 report called "Standards of Ethical Conduct for Employees of the Executive Branch." This is a
30 report that each of you received when you were appointed to the committee. I'm just going to
31 review one of the rules in that document.

32 Before every meeting, you provide us with information about your personal,
33 professional, and financial interests, information that we use to determine whether you have
34 any real, potential or apparent conflicts of interest that could compromise your ability to be
35 objective in giving advice during committee meetings. We waive conflicts of interest for
36 general matters because we believe your ability to be objective will not be affected by your
37 interest in such matters; but we also rely to a great degree on you to be attentive during our
38 meeting to the possibility that an issue will arise that could affect or appear to affect your
39 interest in a specific way.

40 In addition, we have provided each of you with a list of your financial
41 interests and covered relationships that would pose a conflict for you if they became a focal
42 point of committee deliberations. If this happens, we ask that you recuse yourself from the
43 meeting and leave the room.

44 If you have any questions about these rules and your interests, our
45 committee management officers can help address those questions. David Alperin and Claire

1 Harris are this committee's committee management officers, and David at least is here today,
2 and I think Claire will be here later.

3 Thank you.

4 DR. WILLARD: Ed, could I ask Sarah a question on that issue?

5 Could you address explicitly how we are supposed to behave in discussing
6 issues in which either we or colleagues at our institutions either have grant applications in to
7 the federal government or are anticipating those applications? So as we prioritize issues, there
8 is at least an indirect potential for conflict of interest in leaning the deck in areas where we
9 have our own expertise or institutional expertise.

10 MS. CARR: I would still say those are general matters. The discussions
11 that we have about those things, and if the committee decided to make something a priority
12 where you are doing research -- let's say pharmacogenomics -- the fact that the committee made
13 that a high priority isn't going to, in my estimation -- and we can talk to our committee
14 management officers about this more specifically. But in my estimation, you would not be
15 favoring yourself in any specific way. So as long as the discussions are about general issues
16 and you will not benefit specifically from decisions of the committee, I think you'll be okay.

17 But if you ever have any doubt about it, and I know what you're trying to do
18 is get clarification, but if you have any doubt, just come up to me and we'll try to take care of it.
19 I think if you ever have a doubt, the best thing to do is to leave.

20 David, go ahead.

21 MR. ALPERIN: Hi. I'm David Alperin with NCI's committee management
22 office. We do all your ethics clearance.

23 In relation to your question, the specifics of ethics clearance is for you as an
24 individual and the holdings that you have that you would receive a direct benefit if you had
25 insider knowledge of a decision to be made by this committee. If your colleagues have grant
26 applications, that is not a specific issue for you. That is an issue for your institution, and you
27 are not here representing your institution. You are in here for your specific professional
28 expertise. So I don't feel that anything related to a colleague that may or may not have a grant
29 application in process or a grant application for your institution would be an issue that would
30 require you as an individual to recuse yourself from the discussions.

31 Does that help?

32 DR. WILLARD: Sort of. I mean, I guess the issue, without hammering this
33 too deeply, is if I had a grant application in on genetic non-discrimination, it seems to me to be
34 in my best interest to have this committee decide that genetic non-discrimination is one of the
35 big issues that we want to have the Secretary put his weight behind. So would you consider
36 that to be a direct conflict or not a direct conflict?

37 MR. ALPERIN: I would say that that is not a direct conflict. However, it
38 may be a potential conflict. If this becomes an issue of a very focussed discussion, it would
39 probably be the best course of action for you to recuse yourself from that specific section of the
40 discussion.

41 DR. WILLARD: Okay. Thank you.

42 DR. McCABE: The other thing that I will remind everyone is that as a
43 special federal employee, you should not engage in any lobbying of the government agencies or
44 on the Hill while you're here on this visit. Similarly, I think it would not be wise to be involved
45 in any discussions with any companies or relevant to any private holdings that one might have

1 involvement with.

2 I think that the discussion that we just had is that if you feel uncomfortable
3 about it, then there's a perception of a conflict of interest and it's probably best to recuse
4 yourself at that point, since it's frequently perception as well as reality.

5 Thank you for that discussion and for pursuing it further.

6 Emily Winn-Deen will now review the process and outcome of the inter-
7 meeting priority-setting project.

8 Emily, let me thank you very much for chairing the task force. I know how
9 much time and effort you put into advancing this during the interim and in the issue
10 identification process and planning and organizing the meeting.

11 Let me also remind the committee that as Emily reviews the process and
12 issues, you should be thinking again about your rankings. We will take a straw vote after
13 Emily's presentation before the break so that Sarah and her staff can have time to tally that vote
14 over the break.

15 Thank you.

16 Emily?

17 DR. WINN-DEEN: Well, first I want to thank everybody who worked on
18 the committee, and that includes all the people who provided input both from our SACGHS
19 committee, the ex officios, and all the staff support that we got. It was really a group effort.

20 As Ed mentioned, these are the individuals from the main committee that
21 participated, but we did have, when we sent out things for votes to the whole committee,
22 everyone had a chance to vote and participate.

23 The goal of the task force was to conduct a systematic identification of
24 issues that might be appropriate for this committee to address, with the goal of prioritizing
25 some very specific issues that we could and should address; and then as a result of that to try
26 and develop an agenda for this meeting, and that work product is the agenda you see before
27 you.

28 It was a multi-step process. We first identified a number of issues, and this
29 came from feedback that was obtained at the very formation of this committee on what issues
30 were of interest to both the ex officios as well as the members. We then reviewed the top ten
31 issues and assessed how to frame those as short phrases. Then we surveyed the members and
32 asked them to pick their top three to five priority issues. So they got a list of 19 issues that
33 were identified as potentially relevant to this committee's work, and then everyone was asked to
34 vote on which they ranked as the top priority.

35 Then Sarah and her staff organized the results of that first vote based on the
36 frequency, and 12 top issues emerged. Then the full committee went out and worked on that.

37 So again, we surveyed all the members and the ex officios to rank the top 12
38 issues, and that guided the development of the position papers or background paper I should
39 say, issue briefs that you find in your binders. We also identified that coverage and
40 reimbursement was a fairly highly ranked issue among both the ex officios and the members of
41 the committee proper, and arranged to have this as one of the sort of deep issues on the agenda
42 for today.

43 So we went through and tallied all the votes, and these votes were from the
44 committee, and this is sort of the result. The top-ranking issue was large population studies and
45 education and training. They both received a total of seven votes. Coverage and

1 reimbursement and access received six votes each. Then the next group was patents and
2 access, nature of genetic information, oversight and public awareness. Next came the vision
3 statement, direct-to-consumer testing, and pharmacogenomics. And then at the bottom -- and
4 these were the issues that we ultimately decided we would drop, with one exception -- were
5 enhancement versus treatment, bioterrorism, new health-related applications, genetic
6 discrimination, scope of genetic technologies, informed consent, forensics, privacy and
7 confidentiality. The one that we kept on the agenda as sort of an ongoing issue that we're
8 monitoring is genetic discrimination.

9 As I mentioned, 11 issues rose to the top, and the genetic issue was retained
10 because we had already identified this at our very first meeting as an issue that we wanted to at
11 least keep some vision on, make sure that things were happening to take care of that. So the
12 bottom seven were dropped and the rest went on to the next round.

13 In the next round we asked the committee and the ex officios to think about
14 the following questions in terms of trying to prioritize these issues. How urgent is the issue?
15 That is, is there some lurking thing that we have to act on right now? Does the issue warrant
16 the committee's attention? Is it an important issue but maybe not really within our scope? Is
17 there other media attention or some event that's happened that precipitated a need for us to react
18 to that? Of course, does the government have jurisdiction and authority? Because it doesn't do
19 us too much good to work on topics which Health and Human Services can't really take action
20 on.

21 Then the other issues are is there actually already some activity in the
22 federal government addressing this issue? Is the issue of a nature such that the best place to
23 address it is through the government, or are there already other agencies and private sector
24 mechanisms happening to take care of that? Are there any particular moral and ethical
25 concerns that warrant government intervention or leadership on an issue? And then finally, it's
26 the "so what" stuff. Will the committee's policy advice on the issue significantly benefit
27 society, or will the failure to address cause some harm? Does there exist a sufficient body of
28 data from which we can actually deliberate and make a recommendation?

29 So again, we surveyed the members and the ex officios, and there were
30 some differences that emerged between what the members thought and what the ex officios
31 thought, and we'll look at that in a minute. Access was ranked first by members but only tenth
32 by ex officios, so there's some difference in priorities or viewpoints there. Coverage and
33 reimbursement was ranked second by the members and ninth by the ex officios. On the other
34 hand, public awareness was ranked seventh by the members but number one by the ex officios.
35 So I think we need, even within our own extended committee, there's some open issues and
36 need for debate about really what are the priorities.

37 So this is the overall ranking, and I'll let you have a look at that. I
38 highlighted the ones where there was sort of significant differences. I think there were not too
39 many where there was obvious ranked number one by both, so we still I think have some
40 sorting out to do at this meeting.

41 What we did then was we asked Sarah and the support staff to develop an
42 issue brief on each of these topics to sort of flesh them out a little bit more. What people were
43 voting on were these one or two or three-word titles. So it was clear that maybe as we had our
44 task force discussion, not everybody's interpretation of what that title meant was quite the same.
45 So we went through to try and really flesh out what we believe the issues are surrounding this

1 word in relationship to the SACGHS charter, and we'll be going through that in a little bit.

2 I'll give you sort of the highlights of each of these areas on the issue
3 statement, and then we'll come back and have some deliberations. Under access, the key issues
4 that were identified were barriers to access to genetic services that might prevent the realization
5 of the full benefit of advances in genetics, and the fact that access can be impeded in several
6 ways. It can be impeded during the test development and marketing process, through the use of
7 intellectual property patents that might be used in a way that would limit research in an area or
8 increase the cost of tests through licensing royalties.

9 In genetic research, the choice of populations that are available to study
10 might impact ultimately the access of individuals who would benefit from testing to that. So if
11 we never do the studies to determine what the genotype/phenotype correlations are, we're not
12 going to have genetic tests that would benefit, and how do we deal with the rare disease issues.

13 The issue of clinical integration, how do we move from a research level into
14 the practice of medicine; and finally, the sort of standard players in terms of financial barriers,
15 lack of insurance, lack of coverage, inadequate reimbursement, and the cost of the test.

16 From the societal point of view, we also felt that access could be impeded if
17 there was any fear from genetic discrimination or some kind of stigmatization that would create
18 unwillingness for someone, even if it was paid for and available, to have a test, and that this
19 could then, if there are specific groups that feel more likely to be stigmatized, that they might as
20 a group have some disparity in access. Finally, the relevant policy question is are there specific
21 things which the federal government could do to intervene which would minimize the barriers
22 to access.

23 For coverage and reimbursement, we all know that health insurance affects
24 both the cost of the overall health care system and the quality that's delivered, and as a result
25 the access to care. So coverage and reimbursement decisions, particularly for new
26 technologies, new markers, are still very difficult in this country. We're a multiple payer
27 system and we have multiple payer disparities in terms of what's covered, how it's covered,
28 when it's covered, and that all has an impact on who actually is able to get testing done.

29 Some of the coverage and reimbursement decisions are not made because
30 there's felt to be insufficient data to support that something really is medically relevant. There's
31 some misunderstandings about the costs associated with genetic technologies, and there's some
32 new challenges that genetic technologies pose in the paradigm of health insurance, that
33 sometimes it's necessary to test family members in order to get a specific result for a proband,
34 as well as the fact that we now will be potentially able to do testing for diseases that would
35 develop much later in life but are currently asymptomatic.

36 So again, coverage and reimbursement. The policy questions are basically
37 focused on is it a barrier to allowing people to get access to the genetic technologies, what
38 specific actions would facilitate coverage and reimbursement decisions, and are there any
39 unique characteristics that impact these coverage and reimbursement decisions, or are these
40 really just like any new biomarker that comes out and has to establish itself on the basis of
41 clinical utility.

42 We heard at a previous meeting about concerns in direct-to-consumer
43 marketing, and this is basically focused on the marketing of medical services and products to
44 consumers. It's common practice now with all the television and print advertisement for
45 pharmaceuticals, so there's some concern that genetic testing might not be quite ready for that

1 level of consumer interaction. So the risks really are who is regulating the claims that are made
2 in a direct-to-consumer situation, and the potential harm that could be done to this field if the
3 public's first interaction with these kind of tests is through sort of junk science.

4 The average consumer we felt doesn't really have the background and
5 experience to judge for themselves what's good science and what's junk science, and there
6 might be some need for "experts" to weigh in on that. Then there's the whole issue of how
7 many results do you want going directly back to a consumer without the intervention of a health
8 professional, and which kinds of tests might be okay to have results go directly back to a patient
9 and which ones you would definitely want to have a health care professional and potentially a
10 counselor involved with.

11 So the benefits of having some kind of intervention would be to enable the
12 consumers to be better informed and to participate more fully in their health care. We all know
13 that the first thing anybody does today when they're diagnosed with Disease X is log on to the
14 Internet and find out what Disease X is really all about. So in terms of being an informed
15 consumer, that's a good thing. We do have oversight both from FDA and FTC to protect
16 consumers from false and misleading advertisements, but unfortunately right now we've heard
17 from both FDA and FTC that neither agency has the bandwidth on their staff to be actually
18 monitoring this.

19 So the policy questions come down to basically do the risks of direct-to-
20 consumer advertising outweigh its benefits? Does direct-to-consumer advertising in this
21 particular field raise any greater concern or warrant more attention than any other area of
22 medicine? And is there anything that this committee can do to sort of facilitate the right kind of
23 consumer interaction with genetic testing?

24 The next area was genetic discrimination. We've talked about this a fair
25 amount. We know that genetic technologies have been sold on the promise of the future of
26 medicine being positively impacted. We have just completed a really huge publicly financed
27 program to sequence the human genome, and now we're working on the next phase of
28 understanding the variation among individuals in the human genome. But we're not at the point
29 where it has yet been integrated into the practice of medicine in a routine way.

30 So what are the barriers? Is there a fear that patients would not either
31 utilize genetic services or participate in the basic research that's needed to move from a
32 research setting into a medical practice setting?

33 There's still a perception that genetic discrimination exists, although there
34 are actually not that many documented cases, and the ones that have been documented are ones
35 we trot out on a regular basis. So the question is is this a real fear based on facts, or is this a
36 sort of fear of the unknown or fear of worst case scenario and we just haven't seen the worst
37 case scenario?

38 The policy questions are will a federal law, such as the Senate bill that
39 passed last fall, be effective in preventing discrimination? Are there other areas beyond health
40 insurance and employment that we should also be thinking about? There is a moratorium in the
41 U.K., for example, on life insurance, using genetics to make any risk assessment for the
42 purpose of life insurance. But there are also areas, like adoption and immigration policy, that
43 we really haven't discussed or gotten into at all yet. Then again, what further steps should this
44 committee take to deal with this issue?

45 We also heard at the last meeting a lot about the status of genetic education

1 and training. The goal, of course, is to make sure there's a better understanding of the role of
2 genetics in health and disease. We are I think well acquainted with the fact that we need to
3 involve a wide variety of health professionals and make sure that they have the right education
4 and training to facilitate the integration of genetics, and I think currently there is still a
5 perception that health professionals are not sufficiently trained and educated to meet the goals
6 of having this just as a standard part of your medical care.

7 So what are the gaps and how can we fill those gaps? Is there a role that the
8 federal government should play, or is this a role that should be played, for example, by the
9 AMA and other professional groups? So that was the training and education section.

10 Genetic exceptionalism is sort of a philosophical issue. It basically comes
11 down to do we believe that genetic information is different than any other medical information,
12 so should it be treated with some special additional considerations beyond the confidentiality
13 and other things that are in place with which all medical information is treated.

14 The critics say that genetic information is just another part of your overall
15 medical history and that there's no real compelling reason to separate it from other information
16 in your history that might be just as damaging or difficult to deal with, such as HIV status or
17 the fact that you'd had cancer five years ago, those kinds of things. The advocates say that
18 genetic information is unique because it is a unique identifier. It does allow you to, with the
19 exception of identical twins, a unique personal identifier. Because it's inherited, anything that
20 you find out about an individual could have implications for family members, and it can be
21 predictive, and therefore it can be used to stigmatize and discriminate.

22 So this is sort of one of those ongoing debates, whether we want to make it
23 an issue, and what, if anything, this committee can do about coming down on one side or the
24 other, whether we believe genetics is exceptional or not. Clearly, the fact that we have a
25 committee focused on genetics says that we at least think we need to talk about this issue.

26 So in terms of policy questions, I think I already mentioned the issue of
27 does the fact that genetic information is individually unique warrant special attention. Should
28 our public policies be based on the premise of genetic exceptionalism, and is there an
29 alternative concept that would allow the special features of genetic information to be
30 acknowledged without necessarily creating a whole separate and parallel set of rules and
31 regulations for genetics?

32 The next issue is large population studies. This basically is focused on the
33 concept of translational research, that we need to understand better the genetic variability
34 within populations and across populations and the impact that that has on the way individuals
35 develop disease or react to drugs used to treat disease. The federal government has been
36 funding a number of studies aimed at understanding the extent of variability and creating
37 haplotype maps to help us with the research tools. I think we've been doing a good job of
38 funding the research tools, but do we need to do the next step of actually initiating a large
39 population-based study, such as is being done in some of the other countries?

40 One of the reasons to think about this is because for some of the kinds of
41 genetic effects that we might look at in the broad practice of medicine, we are going to see
42 effects that need a large number of people to actually get the statistical power just to properly
43 power the clinical trial, and this might be beyond the realm of individual grant applications or
44 clinical trials that might be funded by a company.

45 So the policy questions really are how important it is to mount large

1 population cohort studies in the U.S., how would we deal with the heterogeneity of the U.S.
2 population. Most genetic studies that have been done in the early phases went to very
3 homogeneous populations to try and find effect. How do you extend that to heterogeneous
4 populations? What should the role of the federal government be, and what obstacles might
5 there be in order to be able to actually conduct a study in the United States where we're a
6 mobile society, we don't have centralized health records? I mean, there's a lot of sort of
7 logistical issues that would come into play that would have to be dealt with.

8 In oversight, we were looking at basically sort of a follow-up of what
9 SACGT did a lot of work on, our predecessor committee, and that is what kind of oversight is
10 needed at the federal level to make sure that the tests that are developed meet all the criteria for
11 being put into medical practice. So basically the issue is have we done enough or are there still
12 some gaps? Clearly, a number of agencies are already actively involved in oversight of both
13 genetic as well as other tests, and the question is is there a need to do more than what they're
14 currently doing? So I guess that's really pretty much those things.

15 Patents and access. This was a pretty hot topic I'd say two years ago, and
16 still I don't think has been completely resolved about what to do about the patent system. The
17 pros, of course, are the basic premise under which the U.S. patent system was created, which
18 was to promote innovation by granting exclusive rights for a limited period of time, basically so
19 that you would invest in developing things and get some kind of assured reward, as market
20 conditions permit.

21 The cons are that when patents are held in an exclusive manner and not
22 promulgated across multiple sites, there's a perception that this can limit innovation or limit the
23 ability to use innovation. Of course, the financial rewards that come from patents are basically
24 through licensing fees. So those who need to take a license and pay fees almost always pass
25 those extra costs directly on to the consumers of their products, including patients who are
26 getting genetic tests.

27 The questions on a more basic level are is there any evidence that patents
28 held in an exclusive manner have limited research or decrease the ability for tests to be out and
29 utilized in terms of access to genetic testing services? So the policy question as it relates to
30 genetics is is there really anything in the way U.S. patent law is interpreted and executed in the
31 field of genetic diagnostics that is unique and requires some kind of special treatment or special
32 consideration, and can we somehow balance the public good that comes from full dissemination
33 of new innovative technologies with the financial rewards that are required to incentivize
34 investors in actually developing those things to the point where they're commercially viable?

35 So again, a balancing act. Is there anything that this committee can or
36 should do to sort of influence how that scale is tipped?

37 In the field of pharmacogenomics, I think it's fairly well recognized that
38 there are individual differences in the way people respond to drugs or are susceptible to
39 disease. There's some evidence that your genetic background also plays a role in your
40 likelihood of developing an adverse event when treated with some drugs. So the question is
41 how to make the best use of this sort of general knowledge and put it into the practice of
42 medicine. So to what extent should we individualize the practice of medicine and use genetic
43 determinants to target pharmaceutical interventions, or to what extent are we using genomics to
44 identify new drug targets and evaluate them in the process of drug development through clinical
45 trials?

1 So the relevant policy questions in pharmacogenomics are basically does
2 the current evidence indicate that genomic technologies can improve health care outcomes?
3 And the part that we haven't really dealt with too much as a society yet is the costs and quality
4 issue. So do we need more health economic modeling of how a test might or might not benefit
5 the overall practice of medicine? How will the clinical validity and utility of pharmacogenetic
6 tests be established? How will pharmaceuticals already on the market be reassessed? We
7 know that there are a number of drugs, for example, that already have in their labeling a
8 statement that says, "By the way, this drug is metabolized by this enzyme, which we know to be
9 polymorphic," but they don't really tell you what to do about that. So how do we develop that
10 body of information that takes it to the next level? And then how will the integration of
11 pharmacogenomics into clinical trials and drug marketing be optimized?

12 So the relevant policy questions in this area are basically what can the
13 federal government do to improve the chance that this technology will be integrated and used to
14 improve patient care? Are there sufficient research studies in place? Is there a role there for
15 the federal government? There's always the fear that if you genetically subdivide diseases into
16 enough groups, that you end up with genetic subsets of disease that might then be sort of
17 orphaned diseases on their own, and do they qualify then somehow for orphan drug status or
18 orphan disease status? And finally, what's the most efficient way to integrate this technology
19 into the health care system?

20 On public awareness, I think we also have identified this and discussed this
21 to some extent at previous committee meetings. The question is just I think focused on how to
22 make informed consumers, how to make sure that the public, and the public being kids in
23 school through elderly on social security, how do we make sure that they have enough
24 knowledge to make good, informed decisions when presented with an opportunity to integrate
25 genetics into their health care, and how to make sure that the public education that occurs
26 through the media is accurate and not misleading.

27 So I guess the key things in terms of what this committee might do is what
28 is the role of the federal government in assuring or improving genetic literacy, and if we can
29 identify what the role is, is there more that the federal government should be doing or could be
30 doing to improve public awareness in the right way?

31 The vision statement issue is really not an issue, more a mechanism to help
32 this committee formulate its framework, and we have a very, very broad charter and basically
33 are given a wide range of things that we could work on. The thought was that if we could
34 create a vision statement, it might help us to describe where we want to get. What is the goal
35 ultimately of integration of genetics into health care and into society? Then once we identify
36 what the ultimate vision is, then you can take a step back and do a gap analysis and say really
37 where are the gaps, what are the most critical gaps, and which ones do we have the ability in
38 this group to influence?

39 So I guess the main policy questions are beyond just giving this committee a
40 chance to get more focused and get a better internal vision for what we want to do, is there
41 some role that such a vision statement could play in becoming a broader Health and Human
42 Services vision statement or a federal government and public vision statement so that we really
43 sort of start with the seed at this committee level and move it up to speak with one voice about
44 where we as a people of the United States want to take this whole area?

45 So that's the overview, and now what we're going to do is we have different

1 committee members assigned to just give us a brief run-down on each of the areas.

2 DR. McCABE: Thank you, Emily.

3 We do have time now for a very brief discussion, probably about 15
4 minutes or so, before we take the straw vote right before the break. So if anybody would like --
5 Francis?

6 DR. COLLINS: Just a point of information. When it came to the
7 discussion about patents and access, you may be interested in knowing the National Academy
8 of Sciences has started a study on that, which met for the first time this past Friday, specifically
9 looking at the impact of patents on genetics, genomics, biotechnology, and a distinguished
10 panel, chaired by Shirley Tilghman, president of Princeton. This will be an 18-month study.
11 They have a lot of expertise on that panel from both the public and the private sectors and will
12 be collecting data in order to try to assess what the impact has been so far of patents in this
13 arena, and then ultimately making recommendations about steps that might be taken to
14 maximize the benefit to the public in the future.

15 DR. McCABE: Thank you. That is important information because, as
16 we've discussed before, there's no point in us taking on issues that are actively being engaged
17 by other groups. There are a lot of things to do, and we shouldn't duplicate efforts. That would
18 not be efficient, nor probably effective.

19 Other comments? Yes, Emily.

20 DR. WINN-DEEN: Francis, can you tell us who would be the right person
21 to contact to maybe have somebody come and just give this committee a briefing when they get
22 to the point of having something?

23 DR. COLLINS: I can get you the name of the chief staff person, and I'm
24 sure they'd be glad to come and make a presentation. They're just getting started with the first
25 meeting, but perhaps in two or three months. I can get you the name.

26 DR. McCABE: Good. Thank you.

27 Other comments? Questions?

28 Yes, Brad?

29 MR. MARGUS: Just on the same lines, I remember at the last meeting
30 someone mentioned that the IOM is looking at the population study idea. I was wondering how
31 much redundancy there would be if we made that an important thing here.

32 DR. McCABE: Francis, can you help us?

33 DR. COLLINS: That has not gotten underway. It's been discussed.
34 Actually, at the present time we're sort of deciding between whether that kind of analysis would
35 be best done in a fashion organized by some of the HHS agencies -- NIH, CDC, and so on -- or
36 whether the IOM would be a useful contributor. We're sort of leaning in the direction of doing
37 that internally. But certainly from my perspective, having the input of this distinguished panel
38 on the value of such a study would be quite helpful.

39 DR. McCABE: Is the IOM still pursuing it, or is the discussion moving
40 toward the agencies?

41 DR. COLLINS: Well, you may be aware, the IOM pursues studies when
42 somebody identifies a budget to support that. So they're looking to the NIH for whether there
43 are funds available. Things are very tight right now, so at the present time the IOM has no
44 plans to initiate such a study.

45 DR. McCABE: Thank you.

1 Other comments or questions? Point of clarification of anything that Emily
2 raised?

3 (No response.)

4 DR. McCABE: Any thoughts about some of the discrepancies between the
5 two lists, the list from the members versus from the ex officios?

6 Yes, Francis?

7 DR. COLLINS: I'm sorry to talk so much, but I'm curious, in the
8 deliberations of the groups that met in-between meetings of SACGHS, there is this apparent
9 potential for overlap between the topic called access and the topic called reimbursement. In
10 fact, reimbursement appears as a subtopic under access. So how did you all come to grips with
11 that in terms of deciding whether these are really different or whether these are two topics
12 where the federal role in terms of what could be done as far as a policy decision is actually
13 fairly similar?

14 DR. McCABE: Emily?

15 DR. WINN-DEEN: It was clear to us that among these 12 topics that you
16 could easily create some subgroups that had common threads, such as access and coverage and
17 reimbursement, whereas you say one of the key issues surrounding coverage and
18 reimbursement is whether it does limit access. But we decided that since they had been ranked
19 as important by enough individuals on their own that we would keep them for now as separate
20 topics. But it is absolutely clear that -- I mean, you could put patents in there as patenting, limit
21 access. So there are subgroups that one could construct and make a sort of coherent subset that
22 might fit together.

23 DR. McCABE: Debra, did you want to make a comment?

24 DR. LEONARD: Well, in fact, there are four of the topics that are
25 subsumed in access. There's discrimination, patents, coverage and reimbursement, and there's a
26 fourth one. I forget what it is.

27 MS. HARRISON: There's large population studies and also --

28 DR. LEONARD: Education. That's the other one that was specifically
29 included in access.

30 DR. McCABE: On one of the conference calls there was some discussion
31 about one of the things that might be done is some lumping, though we wouldn't want to be so
32 creative in our lumping that we ended up not narrowing the field at all, which given the
33 creativity on this committee I'm sure we could probably do. But that would not be effective
34 given the purpose of the process.

35 Barbara?

36 MS. HARRISON: I don't want to get too ahead of myself as the point
37 person for discussion on access, as well as reimbursements. As I was going through
38 information last night, I was trying to separate my thoughts, and it was becoming very difficult.

39 One of the propositions I have for the committee is a topic that was brought
40 up before, if I'm not mistaken by Emily, to use access as a framework for whether or not a topic
41 is worth our consideration or not. It's my bias that in serving the public, to make sure that
42 people can even access these kinds of services, that maybe those are the kind of topics that we
43 need to expand upon, the ones that fit under access. So maybe not identify access as a separate
44 issue but really set it up as a framework in setting these subtopics and these are the ones that
45 we're going to try to address in the next year, couple of years.

1 Again, it may be premature to bring that up, but I wanted to let the others
2 outside of the committee know that that's a thought that's out there.

3 DR. McCABE: I think that was actually Cindy's idea, but thank you for
4 reminding us of that.

5 Suzanne?

6 DR. FEETHAM: Just as a point of clarification, due to a communication in
7 how HRSA scored it, their rankings were not added in, and as the access agency, that may have
8 affected it also. So just to get that on the table.

9 DR. McCABE: As I looked at the discrepancies between the rankings by
10 the members and the ex officios, and this is only my interpretation, but it looked like perhaps
11 what we were seeing was the members just looking and ranking by their feeling of importance,
12 but perhaps the agencies were looking at what could actually be accomplished by the agencies.
13 I don't know if the ex officios would comment on that.

14 Francis?

15 DR. COLLINS: Yes, I think there is some truth to that summary, although
16 that in no way should either imply that the agencies know what they're doing or that the
17 members don't. It's just a different perspective.

18 (Laughter.)

19 DR. McCABE: Yes, Hunt, then Cindy.

20 DR. WILLARD: For the purposes at this point in the conversation, it may
21 be useful to get some guidance before people go into the straw vote, because clearly there are
22 issues of how people read this, or even react to what Emily presented, that we also saw in the
23 task force. So genetic discrimination is a perfect example. Some people rated it number 1
24 because it's a critical issue. Some people rated it number 12 because it's a critical issue, but we
25 already dealt with it because we wrote to the Secretary. So everyone meant the same thing, but
26 the votes were completely opposite, and probably there are six other examples like that. So I
27 don't know how you want to address that from the standpoint of either the first or the second
28 straw vote.

29 DR. McCABE: Well, I think to some extent that will probably come up in
30 the discussion between the first and the second straw vote. But I think it is important to
31 recognize that that probably did influence -- I mean, genetic discrimination was the obvious one
32 that everybody feels is important in the public to SACGT. You know, my first communication
33 as chair of SACGT to both administrations, and our first communication as I was directed by
34 this committee was on genetic discrimination. Clearly, everybody feels it's important.

35 There's the issue of how rampant is it, but the perception is that it's a
36 concern to the public. But I think that will come out. What more can be done about it? If there
37 is more, then that would influence the ranking. But that will probably be a matter of discussion
38 between the first and the second straw votes.

39 Cindy?

40 MS. BERRY: And also, I'd put the genetic exceptionalism in that same
41 category, because you'll remember when we first ranked it and then we looked at it and had a
42 discussion on our conference call, we were all in the same boat in terms of our thinking, but we
43 interpreted our duty to rank it in a different way. So it's the same exact thing, and that one
44 might also merit some pretty intense discussion because it actually has the potential to
45 dramatically affect how we rank our priorities overall and what the committee does.

1 DR. McCABE: Kaytura?

2 DR. FELIX-AARON: Thanks. I just wanted to comment on the apparent
3 discrepancy between the members and ex officios' rankings. It appears that, at least from my
4 perspective, I interpreted the mandate to be, one, what was uniquely the Department's role. So
5 getting clarity and focusing on using that as a priority. So ranking, topics where I thought the
6 Department had a unique role; and two, where action could be taken in the short term. So
7 where there were opportunities for the Department to intervene on a short-term basis. Those
8 were the two things that drove my ranking of the items, and not that the others weren't
9 important. I recognized the importance of others, but I was driven by those two factors.

10 DR. McCABE: Thank you.

11 And I think it's also important to recognize the second point that you made
12 about all of these being important issues. Certainly these are the 12 most important issues that
13 the committee identified. So we recognize in our ranking that we're making some difficult
14 calls, but we also recognize that if we're to be effective we have to have some prioritization. So
15 I don't think we should consider that we don't think that the other points are important. We just
16 have to identify what is the most important where we can be effective.

17 We will also be discussing process, and to the extent that we can come to
18 some closure on one or more of these within this meeting and possibly the next meeting, then
19 we can move on in the prioritization list. But the purpose of this was so that we didn't flounder
20 around not getting our arms around anything, but trying to identify at least three items or three
21 to begin with, and then move on.

22 Debra?

23 DR. LEONARD: There are also things the committee can do that are rather
24 rapid, such as recommending an NIH/CDC study on how we would do a large population
25 cohort that this committee wouldn't necessarily do, and then there are things that this committee
26 would need to do work on. So doing the relative ranking of those types of things is also
27 difficult.

28 DR. McCABE: Thank you.

29 Emily? And I think this will be the last comment, and then we'll take our
30 straw vote and our break.

31 Oh, I'm sorry.

32 MS. MASNY: I was going to say something very similar to what Debra
33 said, that I think if we could clarify what are the important priority issues that we have to
34 address versus those areas where we could make more quick recommendations, because we
35 would want to be able to focus most of our time on those areas where we want to set the
36 priorities rather than spending time where we just need to make a quick recommendation.

37 DR. McCABE: Thank you.

38 Emily?

39 DR. WINN-DEEN: Well, I think we seem to be coming to a little bit of a
40 consensus here in terms of what we might want to do, so I'm just going to try to reiterate for a
41 moment what I think I've heard in this discussion. That is, when we vote this time on priorities,
42 I think we should try and consider what issues are sort of burning issues, what issues the
43 committee can actually do something about in the time frame that we have to work with it, and
44 what are the issues that we believe are important but are either already dealt with or someone
45 else is dealing with them in sort of an active mode. I mean, the goal of this exercise is to focus

1 on getting to a small subset, and we'll say three just as a guiding number, of things that we're
2 actually going to specifically work on and push to take action, not that all of these things aren't
3 important but to try and have some guidance on priority, sort of the "so what?" rule.

4 So if we do something with it, so what? That's just sort of my comment on
5 maybe how we could think about prioritization.

6 DR. McCABE: Okay. So with that, we will vote. It is the blue sheet in
7 your folder. The green is for the second straw vote. Please vote. Recognize that there was a
8 lot of discussion about this on the conference call in terms of prioritization. They're just
9 alphabetical here. That's the way they are listed here. There is no priority intended. It is
10 merely alphabetical on the blue sheet.

11 Please vote, and then Sarah and the staff will pick up your votes and tally
12 them during the break. We will resume at five after 10:00. For members, ex officios and
13 presenters, there are refreshments here. For the members of the public, there is a gift shop out
14 in the lobby of the hotel for refreshments.

15 We'll resume at five after 10:00. Thank you.

16 (Recess.)

17 DR. McCABE: Let's go ahead and get started, then. First of all, I want to
18 just comment on the task force and what you will see is some very fancy footwork now,
19 because each of the members of the task force had topics that they were responsible for. We
20 did not know which were going to be prioritized, so they have all been prepared to lead the
21 discussion.

22 Before we start to look at the screen, as everybody is watching and not
23 really paying attention to my comments, but I just wanted to remind you that really the straw
24 vote will organize the discussion, but there will be another straw vote that occurs later.
25 Remember, the purpose of our discussion today is to help us think through the issues. It's not to
26 resolve the issues, though with a subsequent discussion we're having there may be some that
27 can be resolved very quickly.

28 We're not trying necessarily to address the substance of the issues, and we
29 should not be endeavoring to resolve them, as I said. We want to discuss them in sufficient
30 depth to allow us to weigh the relative significance, and again, problem solving will begin
31 tomorrow, not necessarily today, though with some comments that Chris Hook is going to
32 make, there may be some resolution that could occur to some of these today if the committee
33 wishes to do so.

34 So can we go to the next slide, then? Chris had some ideas that some of
35 these are yes/no. If we look back at the points, the guiding questions, Round 2, it's on page 2 of
36 the handout, the PowerPoint from Emily's presentation. There are four of these points that
37 Chris thought are probably fairly straightforward yes/no and that, in fact, some could be
38 resolved fairly quickly, moved up and dealt with, or moved down and assist us with decreasing
39 their priority.

40 So, Chris, do you want to comment on this, please?

41 DR. HOOK: Thank you, Ed.

42 My concern was that all of these issues are important or they wouldn't have
43 made it onto the final list, and I think it's important that we acknowledge that and that
44 subsequent rankings don't diminish the importance of a given issue by our choices. But we also
45 have to recognize on a practical basis that the Tuckson questions are yes/no sorts of issues. Is

1 someone else working on it? Do we have jurisdiction in the first place? I think we should
2 answer those simple questions first just to acknowledge or recognize those areas where we may
3 be able to move on quickly and say it's not that we don't think the issue is important, but for
4 these simple reasons, practically, we probably should move on to something else. So those
5 were the four that I picked out of his list that seemed to be the easiest to answer in a straight yes
6 or no fashion.

7 DR. McCABE: Thank you.

8 So everybody can see this, and just to comment -- I'll be saying more about
9 this tomorrow -- but Reed is not here today. That's because Reed was picked to be a member of
10 a blue ribbon panel that the NIH has looking at their conflict of interest policy, and that panel is
11 meeting exactly at the same time as we are. But Reed will break away for a couple of hours
12 tomorrow to join us because he has some additional rules that will be important for us to
13 discuss. But I'll talk about those more tomorrow when Reed is here with us.

14 So basically, these are the four points that Chris picked out and mentioned
15 briefly. Does the government have jurisdictional authority over the issue? Does the issue raise
16 concerns that only the government can address, or would the government involvement be
17 duplicative? We heard today about, for instance, the patent issue may fall into the second. Is
18 there another body addressing the issue or better equipped to address the issue? Again, the
19 same thing with patents. Have the policy solutions to the issue already been worked out?

20 So with that, any discussion of these four points? Do people agree that
21 these are fairly straightforward and to some extent yes/no, or at least we can debate the yes/no
22 nature of them on a point by point basis, issue by issue basis?

23 DR. WILLARD: Yes, I agree with Chris totally. I think at the end of the
24 discussion, as you're leading up to straw vote 2, it may actually be better to vote on each of the
25 12 and put them into one of three categories. One is it's not a high priority, so we're not
26 interested in dealing with that at all. A second category that says it is a high priority. We
27 simply want to make a very short statement and pass that on to whoever else is better equipped
28 to deal with it, but at least go on the record as saying it's high priority and this is what we
29 recommend. And then the third class, the relatively small number of issues that are meaty and
30 chewy and that we're going to really get into over the next year, and one of those, just to tip my
31 hat -- I mean, genetic discrimination may be one of those second class issues, the second of the
32 three classes --

33 DR. McCABE: A Class 2 issue.

34 DR. WILLARD: -- because I am very nervous about this committee not
35 saying that genetic discrimination is absolutely a critical, burning issue for us. If we rank that
36 number 10 out of 12, you can bet that will come back to haunt us someday, because the House
37 in particular will look at it and say clearly this is not a burning issue or they would have
38 addressed it number 1. Yet that's an issue that we in a half-hour could draft a quick statement,
39 get it out, and then move on.

40 DR. McCABE: Yes, Joan?

41 DR. REEDE: Speaking against this issue, the genetic discrimination, I
42 actually think among the 12, when we get through with this, there are some issues that are
43 recurring, and when I think about something like genetic discrimination, it could be addressed
44 in all of the other 11 issues that are mentioned. It should be a part of them. I think to pull out
45 some of them that are overarching issues such that no matter what we did a component would

1 have to address something like genetic discrimination -- for example, if it was determined that a
2 population study was important, clearly genetic discrimination would have to be a part of that.
3 I think what that does, it stresses even more the importance. It's saying it's not something you
4 can pull out, examine once and be done with, but rather it's a recurring theme, that no matter
5 what area we go into, we have to look at what is the impact with regard to genetic
6 discrimination. There may be a few others that fall into that same category of being so
7 overarching that we need to look at them no matter what the topic.

8 DR. McCABE: Thank you.

9 With that, and just to reiterate Hunt's categories the way I have them down,
10 number 1 was not high -- and I would say not high enough priority rather than not high priority.
11 Number 2 is high enough priority but can be dealt with briefly, quickly. And number 3 was
12 high enough priority and meaty enough to warrant further discussion. I think those are
13 important points.

14 But with that background, now we will begin an in-depth discussion of the
15 12 issues with the highest ranked issue, which is access. Can we go back to the other slide? So
16 here we can see that we have the members list on the left again, the ex officios on the right. We
17 see that access was number 1 on the members' list, number 3 on the ex officios' list. Some of
18 them moved up or moved down. Some of them moved sort of in both directions on the two
19 lists, up on one, down on the other, to bring them a little closer together. I think it will be
20 important to discuss some of these differences as we go through.

21 Having said that access is the number 1 on the members list, then I think, as
22 I mentioned before, each of the members of the task force was prepared to now be called to
23 action to begin to discuss these issues, and they will serve as our discussion leaders. The task
24 force members will use the policy considerations outlined in the issue briefs with the expansion
25 on those as we just heard. We want to keep the priority-setting criteria in mind. Emily
26 reviewed these before, and as we begin to discuss the specific issues, I think we can go back to
27 -- is it possible to put all of the priority-setting points on the screen? Would that be helpful to
28 everyone? We've sort of separated them out now, so it may be hard. You have them accessible
29 to you in the handout that represents Emily's PowerPoint.

30 So, Barbara, you will be the discussant for access.

31 MS. HARRISON: Okay. I think I just want to very, very quickly
32 summarize the issue brief and explain again that within the issue brief of access, that there are
33 several different subcategories within that that are also on our issue list. These included, as far
34 as the development of genetic technologies, the importance of large population studies that
35 involved diverse populations and pharmacogenetics, and the further development of those
36 technologies, patents and oversight, and the accessibility of the public, coverage and
37 reimbursement, public awareness and education of health professionals, and then finally
38 discrimination so that people do not feel inhibited to pursue such technologies and fear that
39 they will be stigmatized.

40 So I think we will just go right to the questions. I don't want to take more
41 time than necessary. So the first question we have is does the government have jurisdiction or
42 authority over this issue? I can field responses. Any thoughts about that?

43 DR. McCABE: So, access. Does the government have authority,
44 jurisdiction over this?

45 I'll lead off the discussion, and I would say yes, to some extent. Does

1 anybody wish to comment?

2 MS. HARRISON: I guess I could further say that I think because access
3 covers so many different topics, it's maybe difficult to even answer this question because some
4 aspects we may feel government does have control over, while other aspects it does not.

5 DR. McCABE: Yes, David?

6 DR. FEIGAL: Just a quick comment. I think the access falls down into two
7 categories. One is the regulatory category of how things come to market and move from being
8 investigational and access to investigational tests. Some of that's government, but some is also
9 local IRBs, which aren't government. Then there's the CLIA standards and the FDA standards
10 and how they control access. Then there's the indirect government role in access that relates to
11 coverage decisions that practically affects access by determining what you can actually get paid
12 for.

13 MS. HARRISON: Okay, both of which we can have an effect on. So let's
14 move on to the next question.

15 Does the issue raise concerns that only the government can address, or
16 would government involvement be duplicative? Are these issues that if we put forth effort into
17 researching them are things that other people are doing anyway, and so we'd be reinventing the
18 wheel and we could better extend our efforts elsewhere?

19 Joan?

20 DR. REEDE: I think the answer I would have here falls under the same
21 category as the last statement in that there are some things that the government really would be
22 involved in addressing, such as the regulatory issues, the things that are under FDA, et cetera;
23 and then there are some others that may fall under state or federal. I mean, I think it's broad in
24 terms of the response here. So there's a general answer of yes, but it depends on what aspect of
25 access you're looking at.

26 MS. HARRISON: Chris, did you have a comment?

27 DR. McCABE: Please use your mike just so that it can get on the record.

28 MS. HARRISON: Suzanne?

29 DR. FEETHAM: It's also important, I think, to frame this in thinking of the
30 federal role as just by having voice and recognition of it, it may move these programs along,
31 and I think that's a very important component of that. It's not duplicative. It's complementary
32 in the light of its acknowledging the interest of the government.

33 MS. HARRISON: Brad?

34 MR. MARGUS: I absolutely think access is important, and I think this
35 committee's composition, the people on this committee are able to give some really good
36 insights to it. My problem is it seems like a very different word or a different category than
37 everything else on our list because access is going to be discussed in every other area that we're
38 going to vote on anyway. Then if we voted on this one, and let's say we were only going to do
39 access and nothing else, based on what we just heard, then we're going to talk about all the
40 things on the list. It's kind of a different category than the other categories. It's absolutely
41 important, but it's not specific enough that when you say you're in favor of making access an
42 important priority, that you really know what you're going to be talking about because there are
43 so many things it covers.

44 So my vote is that as a category of one of the things to focus on for the
45 committee, I would say we shouldn't make access really one of them. But when we pick our

1 three or whatever number of things we're going to really focus on, make sure access is clearly
2 dealt with when we talk about them.

3 MS. HARRISON: Paul?

4 MR. MILLER: In thinking about the guiding questions, which I found
5 really helpful, I read that a little differently than what's been put on the table. Instead of asking
6 the question does the government have jurisdiction, my sense is this is an advisory committee
7 to the Secretary of HHS, and so in a sense I'm reading it a little bit more narrowly and saying,
8 well, does HHS have jurisdiction. If this committee is coming up with recommendations to the
9 Secretary of HHS but ultimately HHS has nothing to do with FTC issues or patenting issues,
10 then the government may have jurisdiction about it, but it may just not be appropriate for an
11 HHS committee. That's the way that I've sort of been prioritizing these things.

12 It may, in a sense, explain more fully why there's a difference between ex
13 officios and public members of the committee. Public members of the committee may think
14 these are really important issues in terms of genetics health and society. The government being
15 the government, the bureaucrats, may say, well, this is really not an HHS issue; this is my
16 agency's issue. Why are we telling Secretary Thompson about that?

17 So if I am wrong in that this is a much broader mandate than just HHS,
18 that's sort of a good point to discuss. If this is really an advisory to the Secretary of HHS, that
19 may help focus some of these issues.

20 MS. HARRISON: Emily?

21 MS. CARR: I was just going to speak to that.

22 MS. HARRISON: Oh. I'm sorry, Sarah.

23 MS. CARR: One thing to consider about that -- Paul, you raised a very
24 important point, and the committee should be aware that its main purpose is to advise the
25 Secretary of Health and Human Services. But there is a reason why all of the other agencies
26 are represented here. So if the committee decided that, let's say, patents was the highest
27 priority issue, and obviously this is an issue that is handled principally by the PTO, and the
28 Department of Commerce does sit on this committee, it could be that we would decide to write
29 to the Secretary and ask the Secretary to recommend to the Secretary of Commerce that
30 something be done.

31 So I think there is a little room. Obviously, unless the Secretary of
32 Commerce wants advice on that issue, I don't think it would be necessarily appropriate for this
33 committee to offer it. But there are other mechanisms, I think, where you can have some effect
34 on issues that are not solely within the purview of the Department of Health and Human
35 Services.

36 MS. HARRISON: Emily?

37 DR. WINN-DEEN: I think when we worked on this as the task force, we
38 actually did have sort of in our heads a narrower discussion, that this was really what can we as
39 Health and Human Services do. But I agree with Sarah that if there's something we see that
40 needs doing, I think we could still refer it. We maybe shouldn't make it one of the things that
41 we're going to deliberate on, but we should still at least be responsible as members of the
42 community and the public making recommendations to our government to draw attention to it.

43 MS. HARRISON: Also, I'd like to add that under the subject of access,
44 there are topics there such as coverage and reimbursement, Medicaid, et cetera, that do
45 definitely fit under HHS. So I think it's valid for us to consider it.

1 Debra?

2 DR. LEONARD: I agree with Brad and with Joan that this is an
3 overarching issue, that we really need to look at the individual components of access and
4 consider whether those are high ranking or not.

5 MS. HARRISON: So perhaps going back to the suggestion, this can offer
6 as a framework, as a way to look at other issues. It may be possible.

7 Cindy?

8 MS. BERRY: So am I safe to assume that the committee's judgment is that
9 access should be afforded to everyone to genetic technologies, that that's a goal that we want to
10 achieve? When we say access, what do we mean? Do we mean that all individuals, regardless
11 of insurance coverage, regardless of race, regardless of socioeconomic background, whatever
12 else, they should have access to these technologies and the health care benefits that they could
13 produce? Is that kind of the statement that we want to make? Is that what we're saying when
14 we rank it number 1?

15 MS. HARRISON: Emily?

16 DR. WINN-DEEN: Well, I guess I would go back to the Constitution, that
17 our goal, our ideal is equality for all. So, yes, from that point of view, the ideal goal is equality
18 for all. Are there gaps between that ideal goal and reality and practicality? Absolutely. So I
19 think that's what we need to address. Maybe I'm just speaking for my personal voice, but I
20 would say that the overall goal, as it is probably with all health care, is equality for everyone.

21 MS. HARRISON: Hunt?

22 DR. WILLARD: Then that goes to the issue of exceptionalism again. So
23 we end up having to focus, within the general area of access, on which are the specific issues
24 where one might want to tackle access to genetic or genomic services or tests different from the
25 general statement that says everyone should have access to everything in the realm of health
26 care? There's a thousand new technologies that people don't currently have open access to.
27 Should we be saying that genetic technology is somehow different than all of the others, or
28 they're just the same issues that have been dealt with and continue to be dealt with both on the
29 political and scientific end?

30 MS. HARRISON: Joan?

31 DR. REEDE: I would like to suggest that at a minimum the committee take
32 the issues of discrimination and access and say that these are issues that are going to have to be
33 covered no matter which topic. As opposed to discussing is access important, be able to come
34 back when we look at the other topics and see to what extent should access be addressed here,
35 to what extent should discrimination be addressed in each one of them, because I think these
36 are so overarching that if we pick them, basically what we're doing is just starting from ground
37 zero again and saying everything. So I think being able to say when we look at a topic what are
38 the areas of discrimination that we need to take into consideration, what are the areas of access
39 that are relevant here, and making sure we look at them is a practical approach around this issue
40 of can we resolve everything under the heading of access.

41 MS. HARRISON: Kaytura?

42 DR. FELIX-AARON: Yes, I have two points that I'd like to make. One is
43 on the issue of access. I struggle with the scope of that issue because I find it's so broad. When
44 we try to sort of operationalize it, like Emily and others did, I find that it's even more
45 problematic, because what I hear in the statement that everybody should have it, because

1 equality is a value that everybody should have access, is that it can be interpreted as the fact
2 that access to genetic technology is a human right; however, health care isn't a human right.

3 So I think we get quickly into trouble when we make such broad statements,
4 and I just think they raise areas of conflict for us as a committee, but also for the Department
5 when we make those types of statements.

6 MS. HARRISON: Robinsue?

7 DR. FROHBOESE: Hi. Good morning. I do think it's possible to use the
8 construct of access, and I agree with those who have suggested having it as the overarching
9 framework guidance for the committee's activities, and having it really more in keeping with a
10 vision statement or the goal that we are trying to achieve. I think certainly we can support
11 equal access as being fully consistent with the Secretary's initiatives around enhancing access
12 to health care, Healthy People 2010. I think the concept of access without answering the
13 ultimate question of whether everyone is entitled to health care is something that we can
14 endorse and should endorse as equal access to opportunities within genetics.

15 MS. HARRISON: Ed?

16 DR. McCABE: Based on this discussion, we've actually added a fourth
17 category to your three, Hunt, and that was added as a third, keeping the high enough priority
18 and meeting topic as the final category but now a new third topic of an overarching framing
19 topic that needs to be considered in the context of each of the other issues, because I think that's
20 what this discussion is really telling us, that some of these are so important that they transcend
21 through all of the other issues.

22 Is that acceptable to the committee, then, as we begin to think about these,
23 to have that fourth category but we're calling it Category 3?

24 MS. HARRISON: Cindy?

25 MS. BERRY: Is there any other issue that falls under that new category,
26 though? Because I was one of the ones early on that was sort of advocating for access as the
27 framework, and I still think that's the way to go, and I posed the other question just a moment
28 ago because I think we should really nail down what it is we're saying when we pick access as
29 number 1. We could be saying everyone is entitled to it and making the big overarching, lofty
30 statement; or we could be saying it's integral to everything, it's the framework under which we
31 consider all these other issues. There may be another way, but I don't know if there's another
32 issue besides access that falls into that category, that new third category.

33 DR. McCABE: I'd just speak to that point before we move on. I think the
34 other point was made that discrimination really is the other of the categories that is integral to
35 so many other things.

36 MS. BERRY: Do we think discrimination, for example in patents, does that
37 have a role in there? Discrimination is one of those issues that we all care about and think is
38 really a top priority, but I'm not sure it fits or is as pervasive in all the other categories.

39 MS. HARRISON: Martin?

40 MR. DANNENFELSER: I guess I have a question on access. By access,
41 are we saying really coverage and reimbursement? Is it an economic issue we're talking about,
42 or is it broader than economics? Are we saying that people have access by virtue of having the
43 economic wherewithal to get these tests, and then if so, it seems that that's basically -- maybe
44 that's a coverage and reimbursement issue. Or is it a broader question as to who will be
45 allowed to have access to this service, and then the question of reimbursement is perhaps an

1 overlapping but not completely all-encompassing issue in that area.

2 I think just briefly on the genetic discrimination, I think that's a different
3 issue in that I think that that relates very much to the individual, basically kind of the privacy of
4 the individual and things of that nature. So I think that's a different issue. I think that's an issue
5 unto itself largely, but to me it sounds like the way we're defining access sounds very similar to
6 a debate, if you will, about coverage and reimbursement.

7 MS. HARRISON: Ed?

8 DR. McCABE: Yes, I agree. I think that access can be included in
9 coverage and reimbursement, and I think that as an individual that's how I saw it early on. But I
10 think, as I've thought about it more, I really do think it is pervasive. I think that in response to
11 Cindy, is genetic discrimination as pervasive as access, I think they're the ying and the yang, as
12 it were. I mean, lack of access by any individual or group becomes discrimination against that
13 individual or group if an individual doesn't have access to this.

14 So that's why those two, I think, are integral to so many of the others. For
15 instance, with patent and licensure, I think we heard in the Secretary's Advisory Committee on
16 Genetic Testing the concern about Canavan disease families, that because of overly restrictive
17 licensure, they felt that they had been discriminated against. Having supported the research
18 with both their samples and their money, they then did not have access to that.

19 So I really do think it can fit, as I thought about it. It fits into all of the
20 other categories.

21 MS. HARRISON: Joan?

22 DR. REEDE: I agree with that. I think the other part is it is such an
23 important issue for the public that it needs to be seen and stated explicitly, just not implicitly.
24 So I think if we're talking about overarching, to be able to say repeatedly as we look at various
25 topics that discrimination is an important aspect. It sends a message that the committee really
26 does take this seriously.

27 MS. HARRISON: Kim?

28 MS. ZELLMER: I think the other aspect of access is not only coverage and
29 reimbursement but I think that physician education plays a large role in it as well, because if
30 you don't have a physician who can diagnose the problem initially, you're not getting access to
31 genetic technologies that may be beneficial as well. So I think that it does apply in a lot of
32 different areas.

33 MS. HARRISON: So I guess true to form, we basically agree that access
34 covers many, many different areas and maybe just needs to be placed in Class 4. I don't know
35 if that's a decision we can come to at this point or if we need to wait to do that.

36 DR. McCABE: Well, certainly we can come to the decision. We can have
37 these typed up so that people can see them, but the four categories again would be not high
38 enough priority to pursue in the initial consideration; number 2 is high enough priority but can
39 be dealt with in a brief statement or some other rapid approach; number 3 is that it's really
40 important to every other topic, integral to each of the topics. So in that sense, it gets taken off
41 the table because it will be included in the discussion of each of the high-priority topics. Then
42 4 would be high enough priority and meaty enough that it deserves substantive deliberation by
43 the committee.

44 MS. HARRISON: Okay, I think Cindy had a comment.

45 MS. BERRY: Just a point of clarification on the genetic discrimination

1 issue because I'm hearing from folks now a broader definition or concept of genetic
2 discrimination. When we initially looked at it or the scope of our letter, it was confined to the
3 legislation which had to do with using genetic information against somebody for purposes of
4 health care and also employment discrimination issues. It wasn't really discussed as broadly as
5 we are now. We can do whatever we want, and I have no objection to the broader
6 interpretation.

7 But talking about other issues, like if someone uses or volunteers their
8 genetic material for research, are they being discriminated against if they can't get the benefit of
9 that, that's a much broader concept of genetic discrimination than what we originally focused
10 on and what we originally ranked, I think. I have no objection to the broader interpretation but
11 just want to put that out on the table because it's different from what we originally looked at.

12 MS. HARRISON: I agree with you, Cindy. I had the same reaction.
13 I'm sorry, Sarah.

14 MS. CARR: What I was going to suggest is that perhaps you could put
15 genetic discrimination into that transcendent category but also place it in the second one, which
16 is that it's a very high-priority issue but can be dealt with rather swiftly, perhaps through
17 another letter to the Secretary, and you can make an overarching statement as well, that it is one
18 of your highest priority concerns.

19 MS. HARRISON: I don't want to get off on the genetic discrimination
20 tangent too much right now, but I think staying on access -- I don't know if we can just take a
21 quick vote to see if that goes into Class 3 -- is it Class 3 or 4? Class 3.

22 DR. McCABE: Is that fitting with the committee, then? So access, then,
23 would fit into that new Class 3, which is pervasive and should be considered in the discussion
24 of each of the other issues or in the deliberations of any issue that rose to the top in the priority.

25 MS. HARRISON: Is there any opposition?

26 DR. McCABE: Let's take a vote, and probably it's a vote of the
27 membership. We can then take a straw vote of the ex officios.

28 How many individuals would agree that access should go into group 3?
29 Can I see a show of hands?

30 (Show of hands.)

31 DR. McCABE: Anyone disagree?

32 (No response.)

33 DR. McCABE: I'm not seeing anyone who is a member who didn't vote, so
34 it appears that that's unanimous. So then access would move into Category 3, which probably
35 explains why it was number one, because everybody saw it as such a pervasive issue that
36 needed to be considered.

37 Okay. Well, that was very quick, because we had planned to have all the
38 discussion through the morning on that. But I think that was easily dispensed with as we got to
39 the heart of what the real issue was. Thank you, Barbara.

40 So the next is coverage and reimbursement, and you're not off the hook,
41 Barbara. You're still on to lead the discussion of coverage and reimbursement.

42 MS. HARRISON: Okay. Now, on this issue I do want to note that we'll be
43 getting more detailed information this afternoon on this specific issue. However, I do
44 appreciate that our purpose now is to see whether or not it's something that we feel we can have
45 an effect on.

1 One thing I want to note that wasn't really highlighted I don't think in the
2 overview this morning is that the Secretary's Advisory Committee on Genetic Testing had
3 actually done some work on this topic, and I would argue that could offer us a springboard for
4 us to continue. They had come up with two reports, "Coverage and Reimbursement for Genetic
5 Education and Counseling Services," and also "Coverage and Reimbursement for Genetic
6 Testing Services." Those reports, I believe, were not completed, but they were in development
7 when that committee was ended. So one thing I want to offer is that we could possibly
8 continue work on that.

9 They had also defined next steps as including drafting a letter to the
10 Secretary expressing their urgent need for data on health on the economic value of genetic
11 services, including genetic testing education, and had also proposed to convene a roundtable
12 with individuals who have a role in and/or are affected by coverage and reimbursement
13 decisions for genetic services as a way to assess the need for policymaking in this area.

14 So I just want to have those things in mind, again, as we go through these
15 questions.

16 So the first would be does the government have jurisdiction and authority
17 over this issue? I would say that we do.

18 Emily?

19 DR. WINN-DEEN: Well, I think the government does to the extent that
20 there is a government insurance system. But there's also a huge private payer insurance system
21 over which Health and Human Services doesn't have much authority. Maybe influence through
22 where they set Medicare and Medicaid reimbursement levels and what criteria they use, but we
23 do have to recognize that there is a third-party payer system out there. I don't know what the
24 ratio is. Maybe someone from CMS knows what the ratio is between private payer and public
25 payer in the U.S. I mean, it's a substantial contributor.

26 DR. REEDE: It's in our handout. There's a table that describes the relative
27 percentages of coverage, and employment-based private insurance is about 60 percent, and a
28 combination of Medicare and Medicaid is about 25 to 30 percent.

29 MS. HARRISON: Right. The combination I think is around 30.

30 DR. McCABE: I think it is important to point out that CMS establishes
31 through Medicare reimbursement schedules, what is frequently picked up by the third-party
32 payer. So while I agree that it's an influence rather than responsibility, but it is an important
33 influence.

34 MS. HARRISON: So I guess there is agreement that we do have some
35 jurisdiction.

36 Oh, I'm sorry. Debra?

37 DR. LEONARD: There are also upstream aspects to this, like billing codes
38 that get created. That's what the reimbursement is set for, and sometimes the billing codes are
39 inadequate to provide information to third-party payers in the area of genetics as to what is
40 being performed, because the CPT, the billing codes are so generic that there's no real test
41 information provided to payers to provide reimbursement.

42 MS. HARRISON: This is a particular issue with the genetic counseling and
43 education portion.

44 So I think we'll move on to the next question. Does the issue raise concerns
45 that only the government can address, or would efforts be duplicative? My feeling, from what I

1 reviewed, is that our input would be very useful. I don't know if anyone has any other views on
2 that.

3 DR. McCABE: And I think your point in the preamble, your discussion that
4 there were documents that had been prepared and were ready to be rolled out by SACGT, so
5 using those as a springboard, perhaps revisiting them now a couple of years later, that they
6 might serve as a way of getting our arms around this issue fairly quickly.

7 MS. HARRISON: Okay. So I guess I can feel agreement on that issue.

8 The next one is, is there another body addressing the issue or better
9 equipped to handle this issue?

10 MS. MASNY: I still had a statement for the last one.

11 MS. HARRISON: I apologize.

12 MS. MASNY: I'm not in your line of sight.

13 Are there issues that are raised by this area of reimbursement? I think it is a
14 big policy area because one of the things that has been brought out in the brief is that Medicaid
15 itself does not cover screening, and that with the translational effect of the Human Genome
16 Project, that much of the genetic technologies and services are going to move hopefully to the
17 area of prevention and health promotion, where disease has not yet been identified.

18 So I think in this whole area of reimbursement issues, we could make a
19 policy statement about the need to look further into actually better funding, and then of course
20 access to prevention technologies.

21 MS. HARRISON: Emily?

22 DR. WINN-DEEN: I agree that this is definitely an area where we could
23 make some very positive influence. There are also a number of professional group
24 organizations that are taking up this issue, and I would say that if we decide it's something that
25 we want to work on, we should also try and incorporate their expertise and work group
26 products as well. AdvaMed I know has got a broader reimbursement overall of the CPT code
27 system level task force working. So in the context of an overhaul of the whole system, I think
28 we certainly could provide input on what things are missing, need to be there for the things that
29 are inadequately described by current codes.

30 I mean, this is going to be a big issue of do we have to go to the House and
31 the Senate every time we want to get a new screening test or predisposition test, because they
32 don't cover any of that under the current system. In fact, just out of curiosity, I wrote to our
33 reimbursement folks when the latest thing came out on cardiovascular screening, and I said,
34 well, what if we had a genetic predisposition test, would this be covered. And they said, well,
35 cholesterol, yes, but probably not ready for genetics.

36 So there's a mechanism with the U.S. Preventive Services Task Force, but
37 that's where you have to go to get a recommendation. Maybe that's another group that would
38 benefit from getting some guidance on integrating genetics into preventive medicine.

39 MS. HARRISON: Ed?

40 DR. McCABE: And Sarah reminded me that one of the conclusions of the
41 SACGT work group was that there needed to be more of an evidence base, there needed to be
42 development of an evidence base for some of these tests and the recommendations for tests.
43 But again, we could visit that, and that might then intersect with the large population studies as
44 well. I think these are not necessarily going to be independent issues as we go forward, and we
45 can explore those intersections.

1 There may be some where there are groups working on them and it would
2 be duplicative for us to do it. There may be issues like this one where there are a number of
3 groups that are working, both federal and professional organizations working on them, and we
4 can serve as a forum to bring those groups together. It sounds like this is one where that might
5 be the case.

6 MS. HARRISON: So the last question as to whether policy solutions on the
7 issue have already been worked out, I think for the fact that we're talking about this, the answer
8 is no. I don't think there's any dissenting opinion about that.

9 (No response.)

10 MS. HARRISON: Okay. So were there other questions, or did we only
11 want to get through these? I think there was another level of questions.

12 (No response.)

13 MS. HARRISON: How urgent is the issue? Is this something that if we
14 didn't address today, or even within the next couple of months, it would not cause much harm,
15 or is this something that we really do need to put effort toward now? My feeling is that this is
16 something that we need to put effort toward now. We need people to be able to access these
17 services, and already people are running into problems.

18 Any thoughts? Emily?

19 DR. WINN-DEEN: I think it's been pretty clearly identified that there are
20 certainly some specific gaps with coding for services and testing modes that are just not there,
21 and you're forced to use some sort of make-it-fit generic code which drives reimbursements. So
22 I would say there's at least that level of need for some work, but I'm also very concerned if
23 there's a sort of global overhaul, as is being discussed, of the whole reimbursement system, that
24 we need to be active participants in that.

25 MS. HARRISON: Joan?

26 DR. REEDE: I think whether it's anticipating a global overhaul of the
27 system or whatever, I think being very proactive about this, so taking a stance on these types of
28 issues early on, as opposed to waiting until there are more tests available, et cetera, and then
29 saying we need to do something about it would be important for the committee, sort of laying a
30 foundation or groundwork in terms of what are the principles that we think should be operating
31 in terms of compensation and reimbursement, what are the target areas that really need to be
32 addressed or attended to.

33 MS. HARRISON: I just want to pause here. One thing that I'm curious
34 about is that amongst the ex officios, this is not ranked quite as highly as it was amongst the
35 SACGHS members. I was just wondering if anyone had any opinion as to why that was, or if
36 you ranked it lower, why that was.

37 (No response.)

38 MS. HARRISON: No one wants to own up?

39 DR. COLLINS: Well, that wasn't true for all the ex officios. NIH ranked
40 this, I'm not sure, either first or second.

41 DR. McCABE: And why was that, Francis?

42 DR. COLLINS: Of the topics in front of the committee for consideration,
43 going through the guiding questions, trying to identify ones where this committee has strength,
44 jurisdiction, access to decisionmakers, and where there's a pressing issue that no other body is
45 taking on, coverage and reimbursement seems to NIH to be very near the top of the list, maybe

1 at the top of the list.

2 MS. HARRISON: Okay.

3 DR. McCABE: But pursuing Barbara's question to the ex officios, because
4 I think that is important, and it's okay to disagree with the members. But it's important for us to
5 understand why there might be that disagreement.

6 DR. FELIX-AARON: Again, it was not because it wasn't important, but
7 what drove me was the unique federal role, or not even the federal role but the unique HHS role
8 in that. I saw it as a complex phenomenon where points of leverage were clearly within the
9 HHS -- that is, Medicaid and Medicare -- but 60 percent of the points of leverage are outside of
10 the private sector. I mean, private insurance companies are regulated by state government, and
11 many of those policies are local policies. So even when you look at Medicare, a lot of the
12 policy coverage decisions are made locally and not at the national level.

13 So the question for me was where are the opportunities for this committee
14 to provide guidance? Where are the immediate opportunities and where are the specific
15 leverage points? I didn't see those opportunities or immediate leverage points.

16 MS. HARRISON: Paul?

17 MR. MILLER: I guess with all due respect, it's for exactly those reasons
18 that I think coverage and reimbursement should be ranked at the top of the list, because I think
19 the government does have an influence in those issues, not that HHS sort of governs the entire
20 marketplace, but because what HHS does in terms of its government programs is tremendously
21 influential and can be very groundbreaking in setting standards and pathbreaking in terms of
22 what it does. So in terms of policymaking, I think for those reasons this is an important issue.
23 At least it was in terms of our ranking of where there is a gap and where the voice of the HHS
24 Secretary ultimately can be influential.

25 MS. HARRISON: Linda?

26 DR. BRADLEY: I would just add that I agree with the concept of applying
27 leverage, but also the issue of does sufficient data about the issue exist. One of the issues for
28 reimbursement is whether the clinical utility has really been established and whether the case
29 can be made that this is something that should be integrated into health care. So I think that's
30 something that needs to be thought about.

31 MS. HARRISON: Cindy?

32 MS. BERRY: So what we might be saying, then, is not that all genetic
33 technologies should be covered and reimbursed, but maybe if our committee addresses this
34 issue it would be to look at and to build on some things that Joan and others have said, maybe
35 establishing some principles or guidelines for, whether it's companies or whether it's federal
36 programs, how they look at genetic technologies and tests and determine whether it's
37 appropriate to cover or reimburse. When does a technology cross that magic threshold that
38 warrants coverage and reimbursement, and adequate reimbursement?

39 DR. FELIX-AARON: I totally agree with you, and from my agency's
40 perspective we'd be particularly interested in decision support for making policy and coverage
41 decisions. I mean, we do support technology assessment. So as part of that mandate, adding to
42 the evidence base that helps policymakers and other decisionmakers determine what should be
43 covered, how much, doing cost-benefit types of analyses. We'd be very pleased to support that
44 type of building of the evidence base.

45 MS. HARRISON: Brad?

1 MR. MARGUS: So at the last meeting, I can remember Debra making
2 some comments with real practical experience about doing tests for people and how this was a
3 major problem and roadblock that had to be solved. Whereas access was something that
4 transcends all these things, I saw coverage and reimbursement as something that's going to be
5 the ultimate roadblock for a lot of these other things. If you succeed with education and
6 training and we have genetic counselors everywhere giving pre-test and counseling, and if we're
7 doing great large population studies and coming up with all kinds of new discoveries about
8 diseases and pharmacogenomics with predictive markers and everyone is aware of it publicly,
9 but in the end the test can't be given because no one will reimburse, that seems like a real
10 problem to me.

11 So it's really clear to me that we've got to deal with this. I guess my
12 skepticism is just that, again, we may deliberate over it at length and get a lot of information.
13 I'm really excited about the presentations this afternoon. I think because of what happened at
14 the last meeting is why we're doing it. But in the end, are we going to just send a letter to the
15 Secretary that says, oh yes, coverage and reimbursement is important to us? If that's all we're
16 going to do, then it doesn't seem worth all that deliberation. If we're going to really get insights
17 about ways to differentiate between what gets reimbursed or something that's really helpful,
18 then I'm in favor of it.

19 I'm curious as to what others' views are on what we can actually accomplish
20 in this committee on coverage and reimbursement.

21 MS. HARRISON: Ed?

22 DR. McCABE: I was going to make very similar comments about how
23 important this is, because I think the marketplace does drive who goes into the field, who stays
24 in the field, as well as the access of the patients to the technology. If no one is paying for it, it's
25 going to be very difficult to have the technology.

26 In terms of what we could do, I think that's what we could explore, where
27 the tipping points are and how we could actually have an impact. Just saying that we need
28 better reimbursement probably isn't going to accomplish it. What is the evidence base that we
29 need? Certainly, it's important to have evidence-based medicine. However, as a pediatrician, I
30 would point out if we practice evidence-based pediatrics, we'd have almost no medicine since
31 that evidence base is just beginning to be accumulated. So we also have to be realistic in terms
32 of what we can demand of the technology as well.

33 MS. HARRISON: Agnes?

34 MS. MASNY: I was going to make a similar point, and also just to say that
35 one of our mandates was also to identify some of the gaps in the research, and as Linda and
36 Kaytura had brought up, it's not only the clinical evidence but also the gaps in the research,
37 especially in the area of the cost-effectiveness and the cost-benefit ratio, and this may be an
38 area that if further research could be done and the private insurers could see the benefit of such
39 testing, then I think they're more likely to pick up the reimbursement. But we would first need
40 to identify the gaps that would need to be addressed, and this would be something we could
41 make a statement about.

42 MS. HARRISON: Joan?

43 DR. REEDE: Just following up on the thoughts that have already been
44 expressed, for me, we're not at this point in time determining what it is we would do or what it
45 is we would say but rather saying is this important enough for us to have it as a priority area for

1 further study. So just sort of a point of clarification on that.

2 MS. HARRISON: Debra?

3 DR. LEONARD: Following up on what Brad said, as well as addressing
4 the urgency issue, if the coverage for genetic tests is not dealt with so that the reimbursement is
5 higher, there won't be laboratories to do this, because right now most genetic testing is done at
6 academic health centers that basically subsidize this testing with better-reimbursed laboratory
7 testing that's available, and almost all molecular pathology, molecular genetics laboratories lose
8 money because of the billing codes that exist and the reimbursement levels that are set for those
9 billing codes.

10 So as genetic testing grows to consume larger and larger portions of the
11 laboratory testing that is done, these laboratories will not be able to be subsidized like they are
12 now. So while the issue is not so urgent today, if we don't anticipate and correct the problem, it
13 will be a horrendous problem in the future.

14 MS. HARRISON: Robinsue?

15 DR. FROHBOESE: It does seem that given the fact that our predecessor
16 committee had already laid the groundwork and actually had a draft report, that this would be a
17 really important project for this committee to pick up on. As an ex officio member representing
18 the Office of the Secretary, I really do think that this report and a letter to the Secretary is good
19 timing. It fits in with the Secretary's overall initiative on looking at uninsured, underinsured,
20 Medicare reform, as well as prevention. So I think it really ties in nicely with a lot of things
21 that are going on in the Department right now.

22 MS. HARRISON: Emily?

23 DR. WINN-DEEN: Just listening around the table, I'd say that what I've
24 heard is that not only do we think this is an issue that has some value and urgency to it, but
25 there's also two areas where it meets the meatiness criteria. One is just simply reforming the
26 current system to have codes for things with which there is already an established clinical
27 utility but we're not able to properly capture the activities that are required to generate a test
28 result. The other is to think about whether this committee could provide a framework guidance
29 document for what is sort of the criteria that all insurers, public and private, should use to
30 determine the clinical utility bar and cost-effectiveness bar.

31 I think if such a guidance document was available, even if all it is is a
32 guidance recommendation, it certainly would help the people who are trying to get tests to that
33 level to have sort of a common set of goals. If I come in with a package that has A, B, C, D,
34 and E, that meets the reimbursement criteria, and I think it would greatly facilitate the design of
35 the right sets of studies and experiments, whether they're large population studies that the NIH
36 funds or individually funded by diagnostic manufacturers or laboratory validation studies for
37 the home brew environment. To try and create some common framework I think would be
38 enormously valuable to the community.

39 I think SACGT tried to do that sort of with setting different levels of lab
40 tests, but they didn't really address what was necessary to make the clinical utility and the cost-
41 effectiveness arguments.

42 MS. HARRISON: Debra?

43 DR. LEONARD: Well, in dealing with one private insurance company on
44 reimbursement issues for all of infectious disease molecular-based testing recently, their
45 criterion is is there a published paper out there, or papers, that basically demonstrate clinical

1 utility, and that's their criterion. So walking in with your own information about clinical utility
2 has no impact on insurers to pay for something. It's the published literature currently.

3 DR. WINN-DEEN: But that's still a clear goal, then. The goal is to get a
4 publication so that you have a publication. But I think if we could create a set of guidelines
5 that -- I mean, this is maybe getting into more of a detailed discussion but just trying to go to
6 the meatiness argument, I think there are some very specific things that this committee could
7 work on developing and trying to set standards that would be helpful to the community as a
8 whole, and to Health and Human Services.

9 MS. HARRISON: Chris, and then Ed.

10 DR. HOOK: I just want to strongly support Emily's comments. I think that
11 that's a very practical and important thing, working with investigators and clinicians trying to
12 determine when tests should come out, and that influences when they should be reimbursed for
13 clinical services.

14 Just a comment to put a little asterisk by this, this will of course be a bit of
15 an overlap when we talk about the regulation and oversight as we consider that later on, are we
16 really covering now in this discussion most of what we were concerned about in that topic.

17 MS. HARRISON: Ed?

18 DR. McCABE: With those comments, and I think there will be points of
19 contact between these different issues, and we can discuss how much contact there is if they're
20 subsumed by versus just touching upon, but that will be for further discussion. So I'm hearing
21 that the consensus of the group -- I'm not hearing anyone saying that this is not something that
22 should stay on the list.

23 I'm going to just ask us to vote not only -- we aren't really prioritizing now,
24 but I think it does help us to categorize these given these categories. So what I'm hearing is,
25 with the discussion of meatiness, that that is a criterion for Category 4. So is that an agreement
26 that this is Category 4? Anybody who wishes to speak to it being in another category, being in
27 two categories?

28 Cindy, did you want to comment?

29 MS. BERRY: It just sounds like we're categorizing hurricanes -- you know,
30 Category 3, Category 4.

31 (Laughter.)

32 MS. BERRY: I put it in Category 4. That's how I vote.

33 DR. McCABE: Okay. So hearing no dissent to that, all in favor of this
34 being in Category 4, say aye.

35 (Chorus of ayes.)

36 DR. McCABE: Any opposed?

37 (No response.)

38 DR. McCABE: Any abstain?

39 (No response.)

40 DR. McCABE: Okay. So that one we'll move to Category 4.

41 Thank you very much, Barbara, for discussing both of those.

42 Number 3, then, is education.

43 Hunt?

44 DR. WILLARD: This is an issue that was ranked, again, higher by
45 members than the ex officios. I think I can briefly summarize the questions before us in the

1 sense that I think everyone acknowledges there's a gap in genetic knowledge of health
2 professionals. To me, as I read through both the issue brief and thought about it and discussed
3 it with people, there are really two questions to be addressed. One, to what extent is this a
4 federal or an HHS issue, as opposed to that of academic and professional societies and other
5 groups? And the second issue is -- and I always hate to come back to genetic exceptionalism,
6 but this is no exception to that.

7 (Laughter.)

8 DR. WILLARD: The question is how any of us would approach a gap in
9 knowledge in genetics any different than continuing medical education and gaps in knowledge
10 of any other late-breaking, fast-moving field in the field of medicine, and how health
11 professionals would deal with the introduction of any new technology where there probably are
12 gaps, and if there were equivalent groups to this, people might be sitting around saying, gee,
13 how can we possibly get people to understand new radiologic tests or new imaging tests or
14 other kinds of laboratory tests. So to me, those are the two particular issues that frame this
15 question to get us to an issue of whether this is a class 1 or class 4 hurricane.

16 But I think first, probably the point to discuss the most is to what extent is
17 this a federal or HHS issue, because if the answer there is no, then that may drive us very
18 quickly. So if there are any who want to address that particular issue. From my standpoint,
19 other than the obvious training issues that the NIH, for example, is involved in, I'm not
20 convinced this is a federal issue. But I'm just speaking as an individual.

21 Joan?

22 DR. REEDE: I think there's another aspect of this when we talk about the
23 education and training. The part that relates to educating those who are in the sort of
24 professional pipeline versus the continuing education of those who are already out in practice.
25 But the other part that I think is important is the diversity within that workforce and the training
26 and who is being trained, and I do think that there are some roles for the government when we
27 start to look at diversity in training, and I've not heard that as part of the discussion.

28 DR. WILLARD: Again, just for a point of information, do you see that as
29 being specific to genetic and genomic testing in this context, or that's a general issue about
30 diversity and workforce training?

31 DR. REEDE: I think it is a general issue, but I also think it is a specific
32 issue to genetics and genomics. It's something that needs to be at least mentioned or addressed.
33 I'm not saying that this is an overarching issue, but there are a lot of implications if you don't
34 look at diversity within that workforce. So it could be anything from NIH and its training to
35 HRSA and the Bureau of Health Professions. There are different ways in which governmental
36 agencies, DHHS agencies are involved in creating a diverse workforce, and one of the
37 questions within that is is there something that those agencies can also do that addresses issues
38 that relate to genetics and genomics.

39 DR. WILLARD: Other issues? Suzanne?

40 DR. FEETHAM: Speaking from the federal perspective, we have identified
41 for the last several years a commitment to the education of all health professionals in genetics.
42 Part of the reason for that is the rapid expansion of the knowledge base, the concern over the
43 traditional time lag which we find with this rapid expansion of knowledge, with direct
44 application to practice that that time lag is not acceptable in any capacity.

45 Another factor in this, as you've identified, is that it is our responsibility and

1 part of the focus of HRSA for the diversity of the workforce and having the right health
2 professionals across the country in the underserved areas. So that's another piece of this.

3 Another aspect of this is that this is reaching all health professionals, all
4 conditions. And that is when you mentioned about other new knowledge, that's another what
5 we see as unique about this, that there's not one condition or a series of conditions or any age
6 group that this is particularly based with. It's everyone, and that's another perspective of why
7 we see this as an important federal role. Also, as you know and already cited, we have strong
8 partnerships with NIH, with CDC. We have done partnerships with AHRQ and some of our
9 funding, because again we see this as a really very important issue.

10 DR. WILLARD: Other comments? Debra?

11 DR. LEONARD: Well, getting at your exceptionalism, it's reinforcing what
12 Suzanne just said, that the anticipated pervasiveness of genetics to all of medicine does make it
13 somewhat exceptional. I was struck by a comment you made at one of our meetings, that it's
14 like we've discovered a new organ, and it's called the human genome. Much of the health care
15 community does not know about that new organ system, and yet it affects every aspect of
16 medicine.

17 So I don't know that the government directly has jurisdiction to educate on a
18 ground level, but I agree with what others have said, that there are many things that the
19 government can do to influence support, push people to create educational programs that will
20 enhance the health care that's delivered when it moves in a more genetic/genomic direction.

21 DR. WILLARD: Chris?

22 DR. HOOK: I think the jurisdiction is split in that residency training
23 programs have to be approved in order to receive Medicare reimbursement and so on. There
24 would be ways in which the government could require training programs to improve or to
25 document genetics or genomic education in their programs, or at least showing how it's
26 integrated in that.

27 We also could put this on the list of things to write letters to the head of the
28 ACGME and the AAMC and others indicating the priority that we believe they should place on
29 including genetics education strongly in the curricula. So even though that may not be under
30 government jurisdiction, there is some way in which I think the committee may be able to have
31 a broader public impact.

32 DR. WILLARD: Francis?

33 DR. COLLINS: I appreciate the discussion, and I think this is an area of
34 great importance. I take the point that there are lots of other areas of medicine that are also
35 moving rapidly where practitioners need information that's up to date about a field that they
36 may not have had much exposure to, but I think this notion that Debra mentions here, the sort
37 of newly-discovered organ, does seem to be kind of the reaction of many providers when faced
38 with the need to become knowledgeable about a field that they've had really almost no exposure
39 to and have imagined as sort of something abstract that they'll never have to deal with.

40 That being said, exactly what the role for the federal government ought to
41 be is something that we've been struggling with now I think for a decade, and I think there are
42 some answers to that, and I think what Suzanne said about HRSA's role is an important one in
43 terms of what the Bureau of Health Professions is doing.

44 The organization that hasn't been mentioned yet that I think takes this
45 challenge on as its major enterprise, if we're talking about other bodies addressing the issue, is

1 the National Coalition for Health Professional Education in Genetics, NCHPEG. NCHPEG has
2 now been around for almost five years. It has, at last count, over 125 professional societies that
3 have joined up to be part of this, representing virtually all of the major specialties and
4 subspecialties of medicine, of nursing, of social work, of dentistry, nurse practitioners,
5 physicians assistants. A long list of those who find themselves in a provider role have become
6 part of this organization.

7 It has obviously a very huge challenge in front of it, to try to achieve some
8 sort of genetic literacy amongst providers in a short period of time, and it only exists now
9 because of initial support from the Robert Wood Johnson Foundation, which has now segued
10 into support from HRSA and NIH as a major effort. It was greatly facilitated in terms of its
11 effectiveness by being co-founded by AMA, the American Nurses Association, and the
12 Genome Institute. So you had the credibility of the AMA and the ANA from the beginning
13 saying this is really important, because I think one of the things we learned is that the
14 government telling practitioners what they're supposed to know and how they're supposed to do
15 what they're doing isn't always all that effective unless their own leadership is also part of that
16 exhortation.

17 I think NCHPEG has achieved that kind of status. They put forward core
18 competencies for all providers, which have been I think very well received and which are being
19 integrated into the educational plans of many of these professional specialties. There's a lot of
20 CME and other types of activities that are being organized, put out on the web, and integrated
21 into ongoing educational efforts for practitioners.

22 So certainly before starting down this pathway, I think it would be very
23 appropriate to look closely at where the gaps that still exist in this very important agenda, and
24 Joan, your point is very well taken in terms of diversity. I think NCHPEG sees that right now
25 as a very high priority. They have a whole working group aiming to address that. They have
26 succeeded in getting many of the major professional organizations that represent minorities to
27 join up and to bring that expertise to the table about how to do something about that issue.

28 So in no way do I mean to say this is taken care of, but I thought it would be
29 good to have this particular set of efforts in front of the group as you try to decide where to
30 place your bets.

31 DR. WILLARD: Well, if you're not arguing that this is taken care of, then
32 the question obviously is what can this committee do either in terms of collecting information
33 and making that available and/or taking some action that would be perceived as being valuable
34 by someone else.

35 DR. COLLINS: One of the things that NCHPEG initially attempted to try
36 to have an influence over but has not had all that much luck is licensure and certification, to try
37 to get more of a focus on genetics expertise in things like the national board exams and things
38 like state licensure for health care providers. That's a very difficult system to try to influence,
39 and I think NCHPEG, while making some efforts in that regard, has primarily decided to focus
40 on generating materials that professional societies would voluntarily integrate into their own
41 educational efforts and not putting so much time and effort into licensure.

42 But that would be an area, I suppose, where this committee, with its reach
43 as a government-connected enterprise, might be able to make some inroads. I don't know how
44 difficult that would be. It might be fairly difficult, but it's a suggestion.

45 DR. McCABE: So what I'm hearing is that while there are other groups that

1 are involved with this, that there still could be an impact of this advisory committee taking on
2 this topic.

3 DR. COLLINS: And again, SACGT had a whole working group that was
4 focused on education. Joann Boughman, who is here in the room, led that enterprise. It would
5 be worth looking back at that activity and what was suggested to try to pick out of that the
6 things which now, a couple of years hence, have not been attended to that were considered by
7 that group as an important part of the next agenda for the future, but again being very careful
8 not to duplicate things that are already very well underway in other quarters.

9 DR. WILLARD: Ed?

10 DR. McCABE: I was just checking with Sarah. So that paper does exist in
11 the SACGT archives. The working group had not come to the point of formulating, or at least
12 having approved recommendations. But again, there would be some background that could
13 give us -- a couple of years old, but could give us a jumping-off place.

14 DR. WILLARD: Suzanne?

15 DR. FEETHAM: To reinforce what Francis was saying about NCHPEG,
16 and also where we see the federal role coming in, is the encouragement and nudge to the
17 interdisciplinary education and training and practice, and that is something that we can have a
18 perspective on -- and I say we as a government -- in ways that individual organizations without
19 some encouragement would not do.

20 DR. WILLARD: Members of the committee reacting to what we've just
21 heard? Since we need to assign at some point priority to this.

22 Joan?

23 DR. REEDE: Just a general statement as we're listening and we're hearing
24 about coverage and reimbursement and education. If there were other areas where a great deal
25 of work has been done by SACGT prior to this, it would be very useful to know that so that we
26 could have that as some sort of basis and foundation for any of the topics that we're looking at.
27 So to the extent that Sarah or others could inform us about prior work or prior documents, I
28 think it would be helpful.

29 DR. WILLARD: Each one of the issue briefs has a section that does, at
30 least briefly, mention what SACGT did or what it might have available.

31 Other comments?

32 DR. REEDE: But it would be nice to see the whole thing, because the
33 comments are very brief in these statements.

34 DR. WILLARD: Yes, correct.

35 Linda?

36 DR. BRADLEY: Yes, I was just going to comment that obviously CDC
37 also considers education a really top priority, both in the sense of workforce development in
38 general and public health, but also specific training in genomics for the health workforce in
39 general, and has a number of projects, I think one of which you may be aware of, the Family
40 History Project, trying to get the concept of a tool to make the taking of a family history
41 something that's really accessible to physicians in practice and assessing that tool and its
42 effectiveness in identifying individuals at risk.

43 I think also the Center for Genomics in Public Health and their activities in
44 training and technical assistance, both in the states and to anyone in the health professions, are
45 pretty active.

1 DR. WILLARD: Thank you.

2 Other comments on this point? Brad?

3 MR. MARGUS: The meeting isn't interesting unless someone disagrees. I
4 ranked this one high. It's important, but I guess I don't feel that convinced that there isn't a lot
5 of redundancy with the previous committee, with all these other organizations. There are so
6 many organizations that know so much about this and lobby or push in so many different ways.
7 I just don't feel convinced that our committee is going to add that much new value or new
8 insight into the issue.

9 Absolutely, education is critical and we want everyone in all of health care
10 to be informed about the latest things in genetics, but I'm not convinced.

11 DR. WILLARD: Barbara?

12 MS. HARRISON: I'm kind of feeling the same as Brad, and I was
13 wondering if it's a possible option to have this be one of the topics that we want to be updated
14 on at each meeting to give people who are in these types of organizations an opportunity to let
15 us know if there is anything that we can do to help, on a regular basis, as opposed to prioritizing
16 it for us as an issue to actively pursue. I just think that that may be a viable option, because I
17 definitely want to be able to offer people help, these groups help if they feel that we can help
18 them, but again, just in the interest of wanting to focus on issues that we can really make a
19 difference on today, I just wonder if this is maybe something we can put on the side but that we
20 definitely want to be updated about.

21 DR. WILLARD: Ed?

22 DR. McCABE: I just want to comment on process, perhaps. While we've
23 been categorizing things, I think it may be premature to utilize Category 1 at this time. I think
24 that is a category that will be utilized at the end of the process rather than this early in the
25 process. Basically, the issue is does it fit into Category 2, which means high priority, can be
26 dealt with fairly quickly. I don't think it transcends all issues. We could probably force it into
27 that, but I haven't heard anybody speaking to that. So we're really talking about a Category 2 or
28 a Category 4 at this time, and I don't see a way of dealing with it quickly given all of the other
29 organizations that have worked on this and tried to come to grips with it and have not fully.

30 So I would think that it still is viable, and I would suggest that we let the
31 prioritization at the end of the day determine the Category 1 versus Category 4. It's really if
32 we're going to have any sidestep into 2 or 3 during this process.

33 DR. WILLARD: I guess, then, my question would be for this group, prior
34 to taking the vote at the end of all of this discussion, can anyone articulate the kinds of specific
35 value added that this group would bring that other groups are not currently or previously
36 dealing with that might raise it in the individual priority list when it comes time to voting?

37 DR. FEETHAM: Sarah just modified number 2 to say "or through
38 monitoring," and that gives just another perspective on that. It's been identified by Barbara in
39 some of the other discussion. It's important to keep it on the screen. I think your comment
40 about are we going to learn more through more in-depth study through this group, but to keep it
41 visible, to keep an eye on it, to track it, to keep it on high visibility, again with the role of the
42 federal agencies, plus with this group, with the adaption of 2, if people go with that. I think
43 that's another way of dealing with this.

44 DR. WILLARD: Yes.

45 Ed?

1 DR. McCABE: And if we do, I just would point out a significant portion of
2 the discussion has to do with diversity of those being educated and trained, and that should be
3 also an area that we should attempt to monitor should we go to 2, because I think that's one of
4 the most important aspects of this.

5 DR. WILLARD: Emily?

6 DR. WINN-DEEN: So I guess I'm going to disagree a little bit with Ed's
7 comment that we couldn't deal with it quickly, because I think in some ways we could deal with
8 it by putting it in Category 2 and saying we endorse the efforts that HRSA and NCHPEG have
9 undertaken, we support them, we think they're doing valuable and important work towards the
10 goal of integrating genetic training into basically all of the health care workforce, and we would
11 be happy to assist if there's something specific we can do, but put it more on their shoulders to
12 come to us with a specific gap which they would like us to address rather than trying to figure
13 out those gaps ourselves.

14 DR. WILLARD: Okay, thank you.

15 Sorry. I apologize. I can't see down the line here.

16 DR. FELIX-AARON: That's okay.

17 I'd just like to draw a connection for the group, that I see provider education
18 being intimately connected and related to direct-to-consumer advertising, and I think it's a
19 connection that in those discussions we've been having we haven't sort of drawn out that
20 particular connection. I'll tell you why I see it. Much of our conversation has focused on really
21 being concerned about direct-to-consumer advertising, and providers represent an important
22 way that we can deal with that in terms of provider education, patients and consumers going to
23 their providers, wanting more information, wanting to support their decisions.

24 I think that provider education is important in that respect, and I just wanted
25 to throw that point out for the committee to reflect on that connection.

26 DR. WILLARD: Thank you for that.

27 Ed, did you have -- Joan first.

28 DR. REEDE: I see sort of three possible paths. There's a part that's a very
29 quick response that Emily has suggested, a statement of endorsement for some of the ongoing
30 efforts. Also, in light of what Barbara has said, monitoring to see what has changed and maybe
31 reissuing statements or taking other directions with time, as changes are made.

32 And then the third part, going to this issue of because we are dealing with it
33 quickly or we're monitoring it does not mean that we can't address issues of education and
34 training as we look at other priority areas.

35 So for me, if I were trying to do a population study and I had a group of
36 providers that were not educated, I can easily see where I would have major issues around
37 discrimination, access, and a lot of other things in terms of my population study. So I think that
38 it can be revisited. I don't think that the categories have to be so isolated that we can't approach
39 these in multiple ways.

40 DR. WILLARD: Other comments?

41 Ed?

42 DR. McCABE: Just to clarify, I think the point was that I hadn't heard
43 anyone speaking to putting it into Category 2, but I would agree that I think that makes a lot of
44 sense. Likewise, I think we heard on the discrimination that things may fall into more than one
45 category depending on how we look at a topic. So I think that that seems like an excellent way

1 to approach it, that certainly it could fit into -- what I'm hearing you say, then, is Category 2, to
2 some extent 3, perhaps not as pervasive as some of the others, but wherever there's an
3 opportunity to include it in other discussions, we should, and then 4, that there may be more to
4 do on this topic. Is that correct?

5 DR. REEDE: Correct.

6 DR. WILLARD: I think we've had a good discussion and framed the issues,
7 and probably this is one where at the end of the day the chips will fall where they may and we
8 can react to that depending on how highly it's ranked by individuals, or by the group rather.

9 DR. McCABE: So at this point, having heard the discussion of the group,
10 do we want to keep it in multiple categories, or do we want to narrow it down any further than
11 2, 3, and 4?

12 (Laughter.)

13 DR. WILLARD: I think we have some obligation to do a little better than
14 that.

15 DR. LEONARD: Well, especially since you said we can't use 1, so we're
16 not categorizing at all, basically. I would see it fitting into Category 2.

17 DR. WILLARD: Do you want to call for a straw vote before a straw vote?

18 DR. McCABE: Any further discussion of Category 2?

19 DR. LEONARD: Well, when we go to our next straw vote, are we going to
20 vote by category? Are we going to rank by category? Because that would seem to be most
21 appropriate. So we need to put all of these in one of the categories or another so that when we
22 do the final straw vote, they're considered by class.

23 DR. McCABE: Certainly, but we're developing the process as we move
24 forward today, so that's certainly an appropriate way to go.

25 Paul?

26 MR. MILLER: Thank you for inviting me to your sausage factory.

27 (Laughter.)

28 MR. MILLER: What might be helpful to me, and maybe to the group, I
29 think the people sort of regard education and training as important, but what would be helpful
30 would maybe be to imagine or maybe spend a moment or two and think out and say, okay, if
31 this was a really high priority, and if we were going to spend the next four months thinking
32 about -- let's say we've dealt with all the other things -- what would this committee do to sort of
33 move the ball forward on it? Would we hold seminars? Would we sort of wag our finger at
34 medical schools? What would we do for this committee to get our hands around education and
35 training?

36 That may sort of help people in thinking about whether it is a number 2
37 issue or a number 4 issue, because I don't know the answer to that other than the government
38 wagging their finger. That's what we do at the EEOC.

39 (Laughter.)

40 DR. WILLARD: Would anyone like to propose what steps we would take
41 if it turned out to be high priority? The default being if no one can come up with something, it
42 isn't a high priority.

43 Francis?

44 DR. COLLINS: Well, I think first you'd want to collect the information
45 about what's really specifically already being done. You'd want to have a NCHPEG executive

1 director come and tell you all the programs that they're currently pursuing, what their timetables
2 are, what their success rates have been so far in achieving their goals. You'd want to look at the
3 SACGT's document to see what their recommendations had been, and then you'd try to figure
4 out, as you were asking a few minutes ago, are there gaps identifiable here where this group has
5 the jurisdiction and where something that could be done that's not already under somebody
6 else's purview.

7 DR. WILLARD: Ed, and then Joan.

8 DR. McCABE: Joan?

9 DR. REEDE: I guess going back to something I said before in terms of
10 point of clarification, I'm not taking an approach now of trying to figure out what is the solution
11 I'm going to offer, the statement I'm going to make at the end, because I think that's too hard
12 without the information. I see this as a process of trying to figure out what are the priority
13 areas that we need more information on, that we want to study in depth, that we may want to
14 act on more quickly.

15 So without that information, it would be very difficult for me to postulate
16 sitting here something that would be evidence-based and reasonable, that this is a step I think
17 should be taken. I see this as more preliminary, saying do we need to do more in this area. So I
18 think using as a criteria can anybody come up with an action item or an action step is
19 premature.

20 DR. WILLARD: While I accept that, if we don't do that, we still end up
21 with 12 high-priority items that we're chewing through.

22 Emily?

23 DR. WINN-DEEN: I think it's pretty clear that we've identified that CDC,
24 HRSA and NCHPEG are making substantial efforts in this area. Although we could hear in-
25 depth what those efforts are, we at least know that we're not alone, that we've put it as an issue
26 that needs to be dealt with, but there are groups that are actively dealing with it. So my
27 question is just sort of to try and get between items 2 and 4 whether we at this instant in time,
28 with our very limited knowledge about exactly what's going on in all those other programs,
29 would say we think other people are substantially handling it and we just want to endorse that
30 and monitor what they're doing, or do we feel like we at least have an obligation to look in-
31 depth at what they're doing and assure ourselves that they're doing all the reasonable things, or
32 if there are gaps, that then we could address the gaps.

33 So to my mind, that's the issue, whether we want to just spend some time at
34 maybe the next committee meeting or maybe one of the subsequent committee meetings, and
35 we already spent some time at a previous meeting, looking at what education and training was
36 going on. Do we feel like we have enough knowledge to say it's a Category 2, or do we need to
37 gain more information, as Francis suggests, so we can decide if it's a Category 4 or a Category
38 2?

39 DR. WILLARD: Agnes?

40 MS. MASNY: I just wanted to sort of have us take a look at the number 2
41 one again and make a suggestion, that as everybody has been talking, since there are
42 government agencies that are already looking at this issue, that maybe we could say, with
43 somebody else making the connection, that the issues of the workforce training could be a
44 subgroup under some of the other issues, like access. Kim mentioned earlier if the health
45 professionals don't know about a test, then the patient won't have access to it.

1 So I think the workforce issues will be handled under something like access,
2 so that if we could look at Category 2 and say is it a high priority but could be dealt with, and
3 we don't have to say quickly but either through endorsements or recommendations, monitoring,
4 or that it would be handled as a subgroup in one of the other topics, then I think it would maybe
5 help us to look at where -- we still see this as high priority, but it is going to be handled as we
6 are addressing some of the other issues or by virtue of an endorsement or recommendation.

7 DR. WILLARD: Other comments?
8 Martha?

9 DR. TURNER: Just a couple of ways of looking at this, and one is a
10 temporal way, and that is that we all agree that it's happening, and that it's happening in the
11 usual way that technologies are introduced into health care. But if we're not satisfied that that
12 is going quickly enough, then the committee perhaps ought to take action.

13 The other thing is, as a user of the policies or information that comes from a
14 group like this, if I see that this is a priority on your list, then when I'm allocating resources for
15 education or trying to squeeze in one more hour of education to a block of the curriculum, then
16 I'm likely to add this. If it's not there, then I won't. So if this committee comes out loud and
17 strong that education is a priority, I think that has a lot of actions that follow as a result of
18 seeing that. So I would continue to identify it as a priority just because its visibility makes a lot
19 of people pay attention and will get us perhaps to our goal more quickly than if we just let it
20 happen on its own.

21 Medicine is not known for its speed in integrating things into practice, or
22 into our education system. So we could use a little help.

23 DR. WILLARD: Thank you for that.
24 Over to you, Mr. Chairman.
25 I'm sorry, Barbara.

26 MS. HARRISON: I just wanted to hopefully make a clarification with
27 myself, that Category 2 is a no-man's land. So putting something in Category 2 means that we
28 think that it is high priority. It's just that there's no real action that we can take as a committee,
29 although we endorse it and think that's very positive. So maybe a letter of endorsement, as well
30 as monitoring, would be the action, as opposed to just monitoring.

31 DR. McCABE: So you would make it and/or rather than or. Okay.

32 I think what we're hearing is a concern that this is an extremely important
33 issue, but nobody is sure quite how one might grapple with it and fear that after a year's
34 deliberations we might end up where we are today, saying yes, we endorse NCHPEG and
35 HRSA and some of the other activities. I think it's important to recognize, however, what was
36 stated before, and that is that we can have influence on other organizations, like the AAMC,
37 ACGME and these sorts of organizations. That can be done either by extensive deliberation or
38 perhaps by monitoring and including that in a more rapid -- I would doubt that there are very
39 many people sitting around this table that would say that education in genetics is not a good
40 thing.

41 So I think one could include it in Category 2 but not relegate it to a place
42 where it just is hanging there with no action taken. One could take immediate action and then
43 monitor to be sure that things are moving forward, and then if further action is warranted in the
44 future could deliberate on that.

45 Suzanne?

1 DR. FEETHAM: Well, as part of your monitoring, at a future date you can
2 bring some of the parties to the table that you've been talking about, key organizations beyond
3 NCHPEG, but the individual professional organizations, and by just bringing them here having
4 them give you some information in addition to further information from the federal partners I
5 think could go a long way to doing what you're talking about.

6 DR. McCABE: Okay. So some of the organizations that we've heard
7 mentioned in the discussion could be organizations to bring to the table at some point in the
8 future.

9 So it's narrowed down to 2 and 4. Does anybody wish to make a proposal
10 as to which we vote on?

11 Chris?

12 DR. HOOK: I would move that we put it in Category 2 for the reasons that
13 have just been discussed. It is a high priority, but I think if we send a statement that we
14 consider this important, that we at every meeting have a presentation from someone on
15 education, we're communicating, we're continuing to act. But on some of these other issues, I
16 think we're going to find there's a far fewer number of individuals or groups that are working on
17 those topics than this one, and I think that's why I wouldn't suggest we put it in 4.

18 DR. McCABE: Do I hear a second on that motion?

19 Yes, Joan?

20 DR. REEDE: A second on that with the provision that we all understand
21 that as we're talking about education, that diversity is a component of that.

22 DR. McCABE: Discussion of this? And we can talk about specific action
23 later under Category 2. Anyone wish to speak to Category 4?

24 (No response.)

25 DR. McCABE: Okay. So not hearing that, all in favor of education and
26 training, with the additional caveats that we've heard, being assigned to Category 2 for later
27 vote, all in favor say aye.

28 (Chorus of ayes.)

29 DR. McCABE: Any opposed?

30 (No response.)

31 DR. McCABE: Any abstain?

32 (No response.)

33 DR. McCABE: Okay. So Category 2 is where it will go, then.

34 Number four is large population studies.

35 Hunt?

36 DR. WILLARD: This is another one in a very similar state, perhaps, in that
37 the premise here is that we know there are a significant number of large population studies
38 going on in other countries with either particular advantages in the design of their health care
39 system and/or the genetic makeup of their population, and there are discussions in this country
40 already underway, led principally at the NIH, to debate both the need for and design of large
41 population studies here.

42 I think, at least in order for me to frame the issues and think about it, it
43 would be useful to hear from you, Francis, sort of an update on this point. To what extent is the
44 NIH digging into this? So we can evaluate whether it's in good hands and we just need to pay
45 attention, or whether in fact there is something we can actually do.

1 DR. COLLINS: Framed in those terms, I'm not quite sure.

2 DR. WILLARD: I have no doubt that it's in good hand.

3 (Laughter.)

4 DR. COLLINS: Well, thanks for asking. I think this is very much a
5 discussion in evolution. As you stated, we are in a circumstance where there are such large-
6 scale longitudinal population cross-section studies either underway or contemplated in quite a
7 few other countries, but not in the United States. There are some 2 million individuals who are
8 currently enrolled in longitudinal cohort studies on various diseases and who are being
9 followed prospectively and on whom DNA has actually already been obtained. So there's a
10 potential there if one could figure out how to put those together into an enterprise that really
11 did cover the range of possible diseases that you'd like to study, and if the consent was
12 acceptable, and if the study design was acceptable -- there's a lot of ifs here -- to perhaps put
13 something together without having to start from scratch, and that's one of the big questions.

14 Is it possible to cobble together things like the Harvard Health Professional
15 Studies, the Women's Health Initiative, NHANES and a whole bunch of other such studies in a
16 fashion that would accomplish this goal? People that I've talked to both see the advantages and
17 are very concerned about the potential there for that just not being workable because of all of
18 these ifs.

19 We did hold a meeting December 1st, 2nd and 3rd to ask a group of very
20 highly qualified geneticists, epidemiologists and environmental experts because, let me be very
21 clear about this, the point is not just to look at the G part, it's also to look at the E part, and
22 particularly to look at the gene/environment interactions that play a role in common disease.
23 So you'd want to design a study that carefully collected environmental exposure data as well as
24 looked at biological materials, like DNA, cells and plasma.

25 So the consensus of the group -- and this was a pretty distinguished group,
26 and they came in I think with some skepticism -- was that such a large-scale cohort study would
27 be extremely valuable. They had no real dissension from that conclusion. They felt also that it
28 would be useful, if possible, to have this across the age range, from childhood to late adulthood,
29 and if you were going to do so, some of the geneticists argued you may as well do this in a
30 family-based approach so that you were covering three or four generations and you had the
31 ability, using the tools that geneticists are familiar with, to be able to test associations and make
32 sure that they're not false positives.

33 There was some discussion about how this might be connected with the
34 National Children's Study, which is an enterprise which has been under discussion now for
35 some three or four years and which is actually congressionally mandated but not
36 congressionally funded, and that is supposed to involve some 100,000 newborns, actually
37 ascertaining at or before the time of conception, and then following those kids up through
38 adolescence, and would there be a way to put that together with a study that also included
39 ascertainment of diseases that occur throughout the lifespan. There were pros and cons
40 expressed about putting these kinds of things together.

41 So the big issues, of course, that arose out of this were numerous. What
42 exactly would be the study design? What kind of power would you have being able to look at
43 gene/environment interactions for diseases? Of which particular incidence? What could you
44 afford to do? Because the costs would be very substantial and would obviously scale as you
45 went into larger and larger populations. What kind of environmental data could you afford to

1 collect? What kind of genetic data and phenotypic data? How would you deal with collecting
2 clinical information, which in this country tends to be rather fragmentary and non-electronic?

3 Could you take advantage of some of the health care systems that have a
4 somewhat better means of collecting that information and not try to do something in a fashion
5 that depended upon individual paper records in order to collect the data that you need? All
6 those were issues that were put forward and not entirely solved.

7 The huge question I think that hangs over all of this is do we as a country
8 have the national will and the resources to mount a study of this magnitude? If this is going to
9 be useful for looking at the common disorders that people I think would most like to collect
10 data on -- diabetes, heart disease, cancer, asthma, hypertension, and so on -- it doesn't look as if
11 you could achieve the kind of power you'd like for much under half a million people, and that
12 would end up being a very expensive undertaking and one which would have a life that would
13 go on for perhaps two or three or more decades. So you really have to count the cost before
14 you plunge in.

15 The cost would be probably substantial enough that without sort of a major
16 effort at a national level to identify this as a program of considerable importance for the future
17 of our nation's health, it would be difficult to do it. I can tell you, frankly, NIH in its current
18 circumstances, particularly in the budget situation that is affecting us this year, next year, and
19 maybe well after that, there would be no way that NIH could mount this on their own, nor do I
20 think we could with our partners at CDC, who were very much a part of this discussion in
21 December. So it would take quite a substantial enterprise in raising consciousness about the
22 importance of this, akin perhaps to the Human Genome Project some 20 years ago, in order to
23 make this a viable option.

24 So where this all stands is there have been a number of small follow-up
25 discussions, but the plan really is now to try to formalize that a bit more by assembling a
26 working group of experts to try to flesh out some of the questions that didn't get answered in
27 this rather brief two-day workshop. But this is still very up in the air. It was mentioned this
28 morning, the possibility of asking the Institute of Medicine to get involved in this, and that has
29 not been ruled out. But I think the concerns there were partly cost, which again is a real issue
30 right now, and partly sort of timing given that IOM studies generally don't happen overnight,
31 and this is a circumstance where if there's going to be some momentum behind this, we
32 probably don't want to have that go on indefinitely or more than it has to.

33 It might be possible with the expertise that exists at the NIH, together with
34 drawing in a lot of outside experts, and there are a lot of people that you'd want to ask opinions
35 about this, to flesh out the basics of a study design over the course of, say, the next six months.
36 But again, all of this is just completely hypothetical without some very major sense that this is
37 a high priority for public health in this country.

38 The arguments to do this in the U.S. and not simply depend upon the studies
39 that are going on in other places are, I think, fairly convincing. If you're really interested in
40 health disparities, and I think that's one of the major arguments for doing this study in the first
41 place, the studies going on in England or Iceland or Estonia or Germany or Japan are not going
42 to address either the very important minority populations in this country or the environmental
43 exposures that are probably quite different here than in other places. So that's a very
44 compelling argument.

45 I also would think that if we're going to set up a study of this sort, and this

1 was endorsed by the people in December, that it ought to be done in a fashion where there is
2 fairly open access to the data by both public and private sources, so that you really had a public
3 data set that the maximum advantage could be taken of. By the way, I think there's a real
4 chance there for this to be funded as a public/private partnership as well, but we haven't
5 explored that very much so far.

6 So I'm a little at a loss to know exactly what to say with regard to the
7 question that I think you're posing in terms of SACGHS at this moment, on March 1st, 2004,
8 what would be an appropriate role to play. I think if this is going to happen, it will take all of
9 the enthusiasm and scientific support and energy of all of the groups that have a stake in such
10 an outcome, and that would certainly include this group. So it would certainly not belong in
11 Category 1 on your list. But whether this is something for this committee to get deeply
12 engaged in right now when things are very much in flux, or whether this is one to pay very
13 close attention to and see how it evolves in the next few months, you might make a case for the
14 latter at the present time.

15 But I would be very interested in the feedback from this group about the
16 course we're currently on. As you can tell, it's very much a work in progress.

17 DR. WILLARD: Thank you, Francis.

18 Comments? Debra?

19 DR. LEONARD: I think it would be absolutely horrendous, having
20 completed the human genome sequence, which also at the time, as you mentioned, was
21 considered to be an impossible task, to have the human genome sequence and not be able to
22 move it into the realization of all the medical benefits that we anticipated coming from this
23 because we don't have the large patient cohorts. Without this, you're not going to move to the
24 next step.

25 So I don't see how this cannot be a priority and not be something that this
26 committee fully endorses and tries to influence whatever you need to be able to move ahead
27 with this.

28 DR. WILLARD: Would anyone like to agree or disagree with that?
29 Chris?

30 DR. HOOK: I certainly agree with that. I look at some of our other topics,
31 such as pharmacogenomics, and really for pharmacogenomics to become practical on a large
32 scale, we're going to have to do large population studies and see if there is a cost-benefit
33 analysis to prevent the 100,000 deaths a year that are attributed to adverse effects of
34 medication. So I think it's very integral to the other things we've said are priorities.

35 DR. WILLARD: Emily?

36 DR. WINN-DEEN: I think there are two questions. One is, is the study
37 designed disease by disease, or is it global with all diseases coming out of it? That's a question
38 for exactly how it would be handled. I think there's absolutely no doubt in my mind that we
39 have to do this as a country that's committed to making all this stuff affect health care.

40 What I wanted to ask Francis is, having run one of the biggest budget, big
41 science programs in the nation, do you have a sense for if this is bigger or smaller or about the
42 same in scale and scope as the Human Genome Project?

43 DR. COLLINS: It's not been fully costed out because so much would
44 depend on exactly what study design is chosen, but it's certainly on that scale in terms of the
45 investment over the course of a 15-year time period. Much of the cost of a longitudinal cohort

1 study, though, hits you up front, because you need to do the accrual phase. You need to get
2 individuals enrolled and collect the clinical information and the biological specimens, and then
3 the monitoring, the follow-up actually tends to be somewhat less expensive on a year-by-year
4 basis.

5 So again, I think it would take a major national priority being set for
6 something of this sort to go forward, something that could not be done, I think, with the
7 existing resources that are available to any of the research agencies represented around the
8 table.

9 As far as your question about the study design, this would definitely be a
10 cross-sectional study that is not focused on any particular disease. It aims to collect
11 information in a population-based sampling strategy and to follow people and see what diseases
12 occur as you go along. Let me say that's a very valuable part, to have this kind of longitudinal
13 study. It enables you to have less biased case ascertainment, so you're not just collecting the
14 most severe cases, as one sometimes does in a case-control study. It does provide you, though,
15 with nested case-control studies for people that really want to drill down into the specifics of a
16 particular disease and find that the phenotypic information that was collected as far as the big
17 study was not sufficient. You can spring out of this lots of case-control studies focused on
18 specific diseases if they're common enough to have enough incident cases during the period of
19 follow-up.

20 The other aspect of this that bears mentioning, of course, is a huge
21 challenge in terms of how you do the informed consent, especially if we're talking about access
22 by lots of researchers to the material. The participants in this study would have to be not really
23 subjects. They would be full partners in this enterprise and would be engaged in an ongoing
24 way. There's no way this would be anonymized. This would be a circumstance where people's
25 clinical information was part of the record. You could try to protect the identity of the
26 individuals, and we could do that I think fairly effectively with various computational means,
27 but this would be a very different kind of study than what some people have contemplated in
28 the past where there's an irretrievable break in the link between the specimens and the person.
29 You want to be able to go back to them quite regularly.

30 The size of this would be something like 30 times the size of Framingham.
31 But if you look at what we've learned from Framingham about cardiovascular disease, I think
32 we'll be kicking ourselves in six or seven years if we haven't started this study, and if we
33 haven't started it very soon.

34 DR. WILLARD: Ellen?

35 DR. FOX: On the question of whether there's another body that's
36 addressing this issue or better equipped to address this issue, I'd mention the Veterans Health
37 Administration, which in many ways is uniquely situated to address this issue. We have
38 roughly 7 million enrolled patients. We have a highly developed and very sophisticated
39 centralized electronic medical records system and, compared to other health care systems, a
40 really stable patient population that's highly diverse, both geographically and ethnically.

41 The Veterans Health Administration has been actively pursuing a proposal
42 for the type of study that we're discussing here, and that has received conceptual approval from
43 the National Board of our organization, and we're sort of in the final stages of working out
44 some of the specifics in terms of the informed consent issues and the privacy issues. But this
45 would involve collection of environmental data linking to the clinical records system and

1 enrolling on the order of more than a million patients. So that is underway.

2 DR. WILLARD: Thank you.

3 DR. COLLINS: Can I just say a word about that in particular? Yes, I'm
4 very pleased to see the VA is taking that kind of leadership, and obviously the availability of
5 that kind of clinical records system is a wonderful asset. We have also heard of other
6 organizations that are very interested in a similar way, in participation in a project of this sort.
7 Kaiser has expressed that, the Mayo Clinic has. The Marshfield Clinic in Wisconsin has
8 already initiated an effort of this sort.

9 All of these have sort of pluses and minuses in terms of exactly what you
10 could accomplish as far as a broad cross-section of age groups, genders, which obviously is a
11 bit of an issue for VA. But I think some combination of taking advantage of those
12 organizations that have this kind of already strong clinical database and a tradition of carrying
13 out excellent research would certainly be on the table of how you would put this study together.

14 DR. WILLARD: Brad?

15 MR. MARGUS: So back to Debra's comment about this being absolutely
16 essential. I think I totally agree that you want to apply the Human Genome Project's product
17 and studies to find associations and to come up with markers or new and novel targets for drug
18 development. Those are critical and they should be accelerated.

19 I don't think -- my sense is that there isn't as much consensus, absolute
20 consensus that the best study design is to have one huge population that you study, first of all
21 because of the sheer cost, whether it will happen, billions and billions of dollars. But there are
22 learned people out there who also think that sometimes it's better to have different studies
23 where you have experts on whatever phenotype you want to study designing at each time, and
24 to design one group upfront could have a lot of risk. So the consensus isn't absolutely there.

25 The other issue, just getting back to the whole thing, is that right now it
26 sounds like it's all a big debate about infrastructure and resources, of course, logistics. I'd like
27 to have much more consensus that the scientific merit is there in having one big group. I mean,
28 today clinical trials are done everyday for drugs, and they don't use one big population. They
29 go pick their populations.

30 The most important thing is I don't think this committee is really the
31 committee that stands in any way prepared to render a decision about the scientific merit or
32 what the best design is. So while it sounds really exciting, I think we have to wait, and I kind
33 of go with the idea of waiting and seeing what happens as far as the follow-up diligence and
34 figuring out from the scientists who are going to spend more than two days looking at this, and
35 that's why I asked about the IOM this morning. If they then said this looks like a thing that has
36 to happen, I understand completely that Francis would love to have us endorse it. I think we've
37 got to worry a little bit about a conflict of interest. I mean, Francis' company is the Human
38 Genome Research Institute would be the one that carries it out.

39 But if, in fact, the logistics and the scientific merit were demonstrated, then
40 at that point I'd like this committee to absolutely start thinking about the ELSI issues, the
41 ethical issues, the access issues, discrimination issues. There are a lot of things that come up,
42 oversight being one of them. But as of today, I kind of urge the committee to maybe, even
43 though we're very interested in this and we feel it's really important, it doesn't seem ready for us
44 to get involved with. Again, I'd put it in Category 2 where we absolutely want to monitor it.
45 Please, please, please keep us posted on what comes out of it. But it doesn't sound like it's

1 ready yet.

2 DR. WILLARD: I've got Ed, and then Joan.

3 DR. McCABE: I wanted to take some of the same information but come to
4 a different conclusion. I think that there probably will be a need for different types of projects,
5 because certainly we need a very large project that's balanced with respect to gender, that
6 includes children so that we can begin to look at some of the environmental influences that
7 affect our health throughout our life course that begin in childhood, and perhaps even before.
8 So it's important to have that kind of study.

9 It's important to look at other, more targeted studies that may get at other
10 aspects more quickly. The chance that a single study will be the final study I think is highly
11 unlikely. At some point I would hope that, probably not in my lifetime certainly, but we would
12 get to the point where everyone is enrolled in these kinds of studies. If we don't really collect
13 data wherever it's opportune to collect those data, we're going to be missing abilities to develop
14 an evidence base.

15 But looking at the models, that's why I would say some of the same things
16 Brad was saying, but come to a different conclusion, and I think that we could serve as a forum
17 to at least discuss what's going on here in the U.S., what's going on around the world, and
18 looking at what are the opportunities given different models. I think to merely monitor is a
19 little bit too passive for me. I'd like us to take a little more active involvement in this.

20 DR. WILLARD: Joan?

21 DR. REEDE: I'm going to go right in-between the two of you.

22 (Laughter.)

23 DR. REEDE: I don't actually think it's the job of our committee to
24 determine the scientific merit or the study design. I think there are much more learned people
25 who will deliberate on that. But I do think that there are things that our committee could look
26 at in terms of equity, in terms of issues such as if we're starting to use convenience data sets,
27 which I would classify things such as Veterans Administration or some of the other data sets
28 that might be Kaiser or other types of things, what happens to the 43 million Americans who
29 are uninsured and don't fall into those categories who may have very different types of
30 environmental factors or other things going on?

31 So starting to be able to raise those kinds of issues to bring them back to the
32 deliberations; being able to say, as we've already mentioned, issues such as access and
33 education and training, oversight, public awareness. If there's not public awareness, will there
34 be a differential impact? I think those are things that our committee can bring to the front in
35 terms of topics for discussion and saying that as a design is implemented or created or thought
36 about, are these areas discussed, are these areas addressed, and ensuring that they're addressed
37 for our committee, but also for the public.

38 DR. WILLARD: Thank you.

39 Emily, and then Ed.

40 DR. WINN-DEEN: I guess I would like to make sure the committee doesn't
41 lose sight of the fact that there might be some value in what I'll still call large population
42 studies but not quite as broad as the ones Francis described, where the endpoints are more
43 immediate in terms of a shorter time frame, where we could really start to get some
44 translational answers and, as such, develop evidence that would allow genetics to move into the
45 practice of medicine in a time frame earlier than whatever, 15 or 20 years from now when we

1 might have a different kind of answer from this kind of huge longitudinal study.

2 So I think we need to have large population studies in that some of the
3 effects that we're looking for are more subtle than the classic monogenic, highly penetrant
4 diseases, but I'm a little concerned about a long time frame before we have anything and a lack
5 of funding for sort of intermediate endpoints and that kind of stuff.

6 DR. WILLARD: Ed?

7 DR. McCABE: And I think that's the point. I was going to comment that I
8 think Joan wasn't that different from what I was trying to say. She just said it much more
9 articulately than I was able to say it. I think there is a role, probably not in terms of scientific
10 merit, but I think in terms of all of the variables that Joan iterated, as well as some additional
11 ones. But I think there is a role for this committee. Remember, it's the Secretary's Advisory
12 Committee on Genetics, Health, and Society, and I think it's looking at those issues that we are
13 chartered to examine, and looking at the different models and how they might approach the
14 issues of equity that are exactly why this committee ought to take on this topic and why I think
15 it's fairly important.

16 DR. WILLARD: Chris, and then Francis.

17 DR. HOOK: Well, I think listening to the different studies, there's Francis'
18 global study, Emily is talking about a variety of other large population-based studies, and we
19 can envision a number of different ones. But I would think that some of the ELSI issues would
20 be common to them all, and what this committee could certainly do would be to focus on those
21 aspects of it and then provide guidance to whatever large population-based study, be it the NIH
22 or be it some other, that would be performed. Those guidelines need to be out there.

23 DR. WILLARD: I've got three backed up here.

24 Francis?

25 DR. COLLINS: I appreciate the input and the comments. I think those are
26 all very on target. Just a point of clarification, because I was a little concerned that from Brad's
27 comment you might have assumed that I was arguing that this longitudinal study would take
28 care of everything. It absolutely will not.

29 So if, for instance, you're looking at a relatively uncommon disorder, or if
30 you're looking at drug responsiveness, unless that drug is taken by an awful lot of people, you're
31 not going to learn much about it from this longitudinal study. What you are going to learn
32 about are disorders that affect something like a half or 1 percent or more of the population. So
33 the point of that comment is to say case-control studies are going to continue to be absolutely
34 bedrock critical for our study of genes and their role in disease, and this in no way should
35 diminish our enthusiasm for mounting those and running them in the most effective possible
36 way, focused on particular diseases where we really need that information, which is most of
37 them.

38 What a longitudinal study does for you, though, is it allows you to look at
39 interactions between diseases because you're looking at everything. It allows you actually to
40 get unbiased information about environmental exposure, which is a huge problem in case-
41 control studies because there is a recall bias that epidemiologists have written many books
42 about, that if you've been affected already with the disease, your recall of environmental
43 exposures is different than somebody who is currently healthy, and a longitudinal study enables
44 you to get past that.

45 A longitudinal study also provides you, because you're collecting biological

1 specimens, with an opportunity to look for biomarkers that were sentinels of disease before the
2 disease actually occurred, and a case-control study where you're only ascertaining people after
3 they've been diagnosed doesn't give you that chance. So there are these various scientific
4 arguments to say that while case-control studies are critical and we should be doing lots of
5 them, they're not going to give you some of the most important answers about
6 gene/environment contributions to disease, and what we really need is both.

7 DR. WILLARD: Debra, and then Brad.

8 DR. LEONARD: Thank you. I was going to make that point. But my other
9 point is that I think this committee, other than monitoring and looking at ELSI considerations as
10 this moves forward, I think that this committee could also make a recommendation that some
11 decision about doing this and how to do it and how to fund it should be made promptly,
12 because it will be over time, and it takes time to get enough people enrolled and to collect
13 enough data and to follow enough longitudinally that you can start using this as a resource, as
14 an effective resource. There will be things that can be done earlier in the development of this
15 type of population, and then there will be things that fall out later. But it needs to be started as
16 soon as possible so that we aren't 10 years in making this happen.

17 So I would say the scientific issues can be addressed, and this committee
18 could say please do whatever needs to be done to support making a decision about doing this
19 and how to do it, and then consider the funding considerations. But basically, if you don't do
20 this, why did we do the Human Genome Project?

21 DR. WILLARD: Brad?

22 MR. MARGUS: So I would be in favor of having this be one of our
23 subjects to cover if the likely result is that we're going to deliberate over whether we're going to
24 make the recommendation that Debra just said. So if we're going to decide whether we're going
25 to endorse doing it or not. That makes sense. But if it's really for us to go ahead and start
26 discussing all the ELSI issues and access and discrimination, all those issues, it seems a little
27 bit like a waste of time to do it now if we don't even know if this project is going to happen. I
28 mean, are they going to find \$5 billion to do it? Maybe by the time you get around to starting it
29 we'll be much more insightful committee members, five years from now or so.

30 So it just seems a little premature to start down that course of discussing all
31 the ELSI things if it's never going to even happen, and Francis can't tell us today that it's going
32 to happen.

33 On the other hand, if we're going to discuss, keeping in mind at the same
34 time all the ELSI things, but we're going to really discuss should we endorse this, should we
35 say there's a need for this, and HHS and the whole government should start finding the money,
36 as Debra suggested, then I think it's worth having as a discussion.

37 DR. LEONARD: The conclusion to my previous statement that this should
38 be a 2 with a letter, with immediate action and not a 4 -- the 4 may come if the study actually
39 happens and gets implemented. That may become a play for SACGHS to look at the ELSI
40 issues associated with this. But I'm putting it in 2.

41 DR. WILLARD: I'm going to call on myself.

42 (Laughter.)

43 DR. WILLARD: Because I thought I was hearing a reasonable consensus
44 that this was a number 4 issue, and people could then vote among the number 4 issues how
45 highly it would rank, because I would argue that, notwithstanding Francis' discussion of his

1 group and where it's going, there's no guarantee. This isn't a situation that it's either the NIH or
2 it doesn't get done. You could imagine a private consortium getting together to decide to do it.
3 You could imagine one wealthy billionaire deciding that he or she wanted to do this. So I think
4 from our standpoint there are a number of issues that we might address, from the standpoint of
5 genetics health and society, which might add value to Francis' deliberations. It might add value
6 to someone else's thought process on exactly how to go about doing this, whether it's public,
7 private, or somewhere in between.

8 Chris, and then Cynthia.

9 DR. HOOK: Well, to comment to Brad and to support Hunt's statement, I
10 agree. I think it is going to happen, and it will be either a collaboration between medical
11 institutions or private industry or something, but to wait and discuss the ELSI issues until the
12 study is up and going is after the fact. It's too late.

13 MR. MARGUS: Not up and going, just so we know something is going to
14 actually happen. It would be really hypothetical to do it today. We don't know that this is
15 going to happen. I mean, where's the \$5 billion going to come from? Are you sure it's going to
16 happen?

17 DR. WILLARD: But there were three or four years of discussions about the
18 Human Genome Project before we were absolutely sure it was going to happen, at least in the
19 way it finally rolled out.

20 MR. MARGUS: That's my vote.

21 DR. WILLARD: I've got Chris and then Cindy.

22 MS. BERRY: Just a question to the scientists really of the group. What I'm
23 hearing, and especially when I listen to Debra, when you speak it almost sounds like this is the
24 equivalent of going to Mars or some sort of massive vision.

25 DR. WILLARD: It's cheaper.

26 (Laughter.)

27 MS. BERRY: But along the lines that this would be a dramatic
28 development to move science forward -- I mean, if you believe in the human genome and the
29 promise of the human genome, it sounds like what you're saying is that in order to realize the
30 benefit of that, we have to have some sort of national commitment to translating what we learn
31 there into actual practice, research and then clinical practice, and it requires some sort of
32 dramatic commitment on the part of the federal government, HHS, as well as the private sector.

33 Correct me if I'm wrong. This is just what I'm gleaning as I'm listening to
34 people. If that is accurate, then it seems to me that this does rise to a level 4 category, where
35 perhaps we can weigh in with a very strong statement and an opinion after in-depth analysis. I
36 mean, I don't know that we can just jump right out and do a letter to the Secretary saying this
37 would be a really good thing to do, and then we check it off because it was in Category 2. I
38 think we have to really learn some more about the justification for it, what it means for
39 medicine, for society, and after taking all the information in, then weighing in with a strong
40 endorsement one way or the other.

41 I could be all wrong, so I ask really my colleagues, who are much better
42 versed in this area than I am.

43 DR. WILLARD: From my perspective, if it's a number 4, then one of the
44 points would be to address exactly that point rather than presuming that we know the answer to
45 a question that we probably don't know the answer to.

1 Ed, you had a --

2 DR. McCABE: I was going to agree with that. I think that to recommend a
3 study of this magnitude, while we agree, I certainly agree that this is required to really fulfill
4 the promise of the Human Genome Project, I think we'd need to look at what is being done
5 around the world and some of the -- I know the British Biobank has been hung up for several
6 years because of the ELSIs there, because of the open nature of the database and that sort of
7 thing. To discuss that I think would be appropriate.

8 I know there are some others who want to comment, but I think we need to
9 either make a decision that this is going to go on somewhat longer, take a break, get our lunch,
10 come back, because it's going to be a working lunch, or we could try and wrap it up and move it
11 to one of the categories fairly quickly, and then move on to the next.

12 DR. WILLARD: I guess I would drive it to that point. If people are
13 comfortable saying this is level 4 and let's then move on to the next one, we could do that. Or
14 just take a straw vote and see how many 2's versus 4's we have, and then you can decide
15 whether we need to discuss this more fully.

16 DR. McCABE: Debra, you wanted to --

17 DR. LEONARD: My only concern with making this a 4 is that this
18 committee meets relatively infrequently, and I would hope that Francis or others for whom this
19 is more of an urgent issue could do the review of is this scientifically warranted, what are the
20 other models that are out there in a more timely fashion. But I may be wrong.

21 DR. McCABE: My guess is that that's already been done as part of the
22 deliberations we've had.

23 DR. COLLINS: It's certainly not done. We've started it. Again, this two
24 and a half day workshop in December was a good start. We have this working group that's
25 getting formed to look at that in more depth, which will have lots of deliberations in the coming
26 months. I would welcome any kind of connection of that enterprise to this committee that you
27 would find would be valuable, and then we could plan a full presentation at the next meeting of
28 SACGHS to see how far have we gotten with this and what are the areas that this committee
29 feels need further attention and would like to get more deeply involved in, however you'd like.

30 DR. McCABE: I would suggest if we did that we also looked at other
31 models as well.

32 DR. COLLINS: And we are as well in this working group. That's part of
33 the intention. So can we depend on the other models that are going on in other parts of the
34 world? Could we build this study off of existing large-scale cohorts that are already underway
35 where DNA samples have already been collected? What would be the possibility of this being
36 done as a private/public partnership? Which looks pretty encouraging.

37 DR. McCABE: I'll take Hunt's comment as a motion for a 4. Do I hear a
38 second to that motion?

39 MR. MARGUS: Second.

40 DR. McCABE: Okay, Brad has seconded to the motion.

41 Any further discussion?

42 (No response.)

43 DR. McCABE: Not that we're holding your lunch hostage or anything to
44 that discussion.

45 (Laughter.)

1 DR. McCABE: Any further discussion? Because we could table this until
 2 after the break.
 3 (No response.)
 4 DR. McCABE: Okay. Seeing the body language around the table, we'll
 5 take it to a vote, then.
 6 All in favor of this as a Category 4, say aye.
 7 (Chorus of ayes.)
 8 DR. McCABE: Any opposed?
 9 (No response.)
 10 DR. McCABE: Any abstain?
 11 (No response.)
 12 DR. McCABE: Okay. With that, let's take a 10-minute break to gather the
 13 lunches. The lunches for the members and the ex officios are outside the door. They have your
 14 names on them. Please pick them up and come back, take a break for 10 minutes, and then
 15 we're going to have a working lunch today.
 16 Thank you.
 17 (Recess.)
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36 AFTERNOON SESSION

(1:00 p.m.)

37 DR. McCABE: Why don't we go ahead and get started? We still have eight
 38 of the 12 to do. We technically have a half hour to do that in, but we want to be sure that
 39 everything has adequate discussion. We have an hour tomorrow that we were going to discuss
 40 process, but in fact we've been doing that as we go along today, so we can slide over into
 41 tomorrow. There was some sentiment that as we got down to the bottom of the list we could
 42 spend less time, but that may be because they haven't had adequate discussion here. So I'm not
 43 sure that we want to do that.
 44 So we may hold our second straw vote until tomorrow after we've discussed
 45 all 12. Does that seem reasonable to everyone? And it's doable, Sarah, with respect to staff

1 needs, to hold the straw vote until tomorrow? Okay.

2 So, Cindy, this is to discuss public awareness.

3 MS. BERRY: And I'll start with just a real general introduction, then my
4 own assessment of how we might consider this issue, and then open it up for discussion to
5 everyone.

6 Public awareness, as you know, is a very broad topic, and what struck me,
7 and it actually was specifically articulated in the issue brief, was public awareness of what? Is
8 it about the need for genetic testing in certain circumstances? Is it the availability of genetic
9 testing? Is it awareness or understanding of results of genetic testing once you get those
10 results? Also other questions that struck me, how much of this is really the responsibility of
11 health care providers versus someone else's responsibility or the federal government's
12 responsibility or HHS' responsibility?

13 Public awareness seems to be more important in the context of direct access
14 to genetic technologies. To the extent that someone is not going through a health care provider
15 or gatekeeper or someone who can really guide the individual, the public awareness and
16 understanding is more critical. Also, if there's insufficient regulation or oversight such that
17 there may be a potential for harm, public awareness becomes much more critical. It's less
18 critical, I think, in the context of an ideal situation where you have educated health care
19 providers who are regularly counseling their patients about the availability of genetic
20 technologies and interpreting those results properly for them.

21 That sort of gets to the question of what is the appropriate role. Is it a
22 health care provider issue? Is there a role for the federal government? Using our four guiding
23 questions, the first is does the government have jurisdiction or authority? I would argue that I
24 don't know that jurisdiction or authority is necessarily the right term here in this context, but
25 certainly the federal government can play a role in educating the public. There are all types of
26 public awareness campaigns that HHS and other agencies engage in. So I think the answer to
27 the first question is probably yes.

28 The answer to the second question, can only the government address this, I
29 don't think that's right, although the government may have a role, and there are government
30 agencies and organizations that are working on this problem. To some extent there may be
31 duplicative efforts in play here.

32 The third question, is there another body addressing the issue, and actually
33 in our issue brief on this topic you can see that there are an awful lot of groups and agencies
34 that are working on certain aspects of public awareness: the Department of Energy, National
35 Human Genome Research Institute, Health Resources Services Administration and the March
36 of Dimes are teaming up, the American Association for the Advancement of Science, and
37 undoubtedly other groups as well. So the answer to number 3 is yes.

38 Four, has there been a policy solution or are there policy solutions that have
39 already been worked out, I would argue no given what we know so far about the level of
40 knowledge of the general public with respect to genetic technologies. So I think we have the
41 possibility of an appropriate government role. There are agencies and private groups working
42 on this issue. We do need some, I think, more solutions because I think there is a problem
43 there.

44 The question is for the committee, how much of a priority do we want to
45 place on this? Is this something that we can and should influence? Do we have concrete

1 recommendations that we would like to consider putting together for the Secretary for
2 additional work, work that's in addition to that which is already being done, or is this something
3 that really is in the Category 1 or 2 where it's somewhat important but there are other issues that
4 are more important, or the second category is it's important but we don't need to spend an
5 enormous amount of time working on in-depth analyses and recommendations. I'm not going to
6 put forward an opinion on that one. I just wanted to kick-start the discussion and then open it
7 up for others, and then we can talk about how we can categorize it.

8 Yes, Agnes?

9 MS. MASNY: Just for clarification, again this is one of the issues where
10 the ex officios had ranked this as the number one issue, and I think your first question about the
11 public awareness of what, that maybe if the ex officios would like to respond to that as to what
12 they saw as the need for public awareness.

13 MS. BERRY: Yes?

14 DR. FEIGAL: I could start. I think that one of the reasons that I thought
15 this was something the committee could do is the committee is in a better position to take
16 advocacy and to take a stand on issues that some of the government positions might have to
17 remain relatively neutral on. Often there's a perception that it's not a good use of public funds
18 for many agencies to have public information campaigns. I'll just give you a small FDA
19 example.

20 It's been debated a lot, although it's finally swung in favor of FDA being an
21 advocate, but for the longest time it was argued that FDA shouldn't have any position on
22 generic drugs, either for them or against them. They should just assure that they were high
23 quality. As the concerns about the economics of health care came up, then FDA got active in
24 pointing out the role that they play in keeping down the costs of prescription drugs.

25 But it's just an example that there are things that can be done in terms of
26 public advocacy, public awareness for these issues that you as the committee can do and can
27 ask the government to do that the government can't ask itself to do.

28 MS. BERRY: David, as sort of a follow-up, do you have any ideas of what
29 you think might be most effective? I know that in the past people have talked about
30 information brochures. I know that there's been some discussion of website activities. Is it just
31 holding forums? Do you have any sense of what the need is, and then what the most
32 appropriate response might be?

33 DR. FEIGAL: Well, particularly because Francis brings examples of some
34 of the nonsense that's out there around genetics from time to time, one of the things that a
35 committee like this can consider is staking its position out as a responsible voice in advocacy
36 for issues around this, whether it's some of the consumer protection or whether it's some of the
37 access issues or reimbursement issues, because you don't have a vested interest or you have
38 some complicated vested interests that no one can figure them out. I'm not sure which of the
39 two it is.

40 But as a body, you can make comments on things. Some of it is even
41 deciding which of these meetings is sort of deliberative and it's fine if they don't have a lot of
42 coverage, as opposed to the meetings where you really have a message that you want to get
43 across and making sure that that message makes it into the press and gets some availability and
44 out there. But there are things like websites and other types of things that certainly would be
45 used.

1 I think the thing that's unfortunate about a lot of commissions and
2 committees like this is that it can be a fairly anonymous task that produces a thoughtful report,
3 and then it just sort of fades from view. I think that if you really wanted to have the ability to
4 make a recommendation that people would point back to, perhaps they wouldn't form the next
5 committee after this one.

6 MS. BERRY: Matthew?

7 MR. DAYNARD: Just a brief comment. I agree with you, David,
8 wholeheartedly. At the Federal Trade Commission, public awareness goes hand in hand with
9 our law enforcement. To the extent you're concerned about consumers being misinformed
10 about genetic testing, what it is, what it can do, what it can't do, whether it's efficacious,
11 whether there should be a doctor involved, you need to get that message out, and we do that in
12 literally every area that we're involved in -- for example, dietary supplements or laser refractive
13 surgery. We've co-authored brochures with the private sector folks, with the FDA. It's
14 imperative that it be done.

15 To the extent that the committee thinks it can do something like that, like
16 David suggested or other things, I think it would be very important for you to do. In terms of
17 websites, we even have something in the dietary supplement area called "teaser" sites.
18 Consumers, as you know, are going on the web as soon as they have a health condition and
19 finding out some good things and some bad things, some accurate things and a whole lot of
20 inaccurate things. We have a site -- it might be a cancer site, I think it's weight loss or arthritis
21 -- and it's a bold claim made on the site, "Order Here," and the consumer keeps clicking, and at
22 the end it says "Gotcha! You would have just lost your income for the last four months if you
23 had bought this product because it's worthless."

24 So if you see claims like this, if you see a genetic test that says we can tell
25 you whether you're going to have a retarded child or something, wrong, you're going to lose.
26 There's a lot that can be done.

27 MS. BERRY: Linda?

28 DR. BRADLEY: I think one of the things that concerns us is the lack of
29 balanced information. I mean, the public is getting their information about genomics largely
30 from the media, and that varies from out-and-out hype to maybe overly optimistic, to sometimes
31 dire predictions. What might be more helpful is for them to have a source of balanced
32 information about what makes sense and what doesn't right now, currently, based on what we
33 know, what are the gaps in information. I've heard Francis speak many times about the loss of
34 credibility that we could suffer if we don't start differentiating between what are tests and other
35 genomic applications that we have some basis for substantiating it should be in practice and
36 others that are being offered -- and we've all seen some of the websites -- where there really
37 aren't data available.

38 So I think that trying to look for that source of a credible, unbiased source
39 of information that we're concerned about.

40 MS. BERRY: Emily?

41 DR. WINN-DEEN: I thought SACGT was working on some kind of a
42 patient or something brochure, and I think it might be helpful if we could also, as you
43 mentioned before, get a copy of that to see what is happening. I guess I interpret patient
44 awareness as much broader, just really making sure that all the education is there through the
45 school years, but I'm particularly concerned about people who have completed school, and

1 there's a lot of them out there, and how are we going to make sure that they're getting correct
2 information and not just reading about it in whatever news source. What's on the Discovery
3 Channel or PBS are probably good programs. There's probably just as many bad sources of
4 information out there, and how do we help people know where to go, what to do.

5 So I'm not sure exactly what this committee can do, but I am really
6 concerned about it as an issue, and I think it does need to stay on the radar screen and be
7 addressed.

8 MS. BERRY: Ed first, then Martin.

9 DR. McCABE: Yes, I was going to comment. There was quite a bit of
10 work that was put into patient information brochure by the SACGT that was never completed.
11 So certainly that is something that could be brought to this committee.

12 MS. BERRY: Martin?

13 MR. DANNENFELSER: It would seem like it would be difficult for the
14 committee to do this on an ongoing basis. Just I think the basic charge of the committee is to
15 be advisory to the Secretary. So most of the ongoing work I think would have to be done by the
16 Department. But you've got a lot of resources within the Department, and then through other
17 agencies. But I think certainly this kind of objective information, because there are a lot of
18 claims being made out there, and they're conflicting, and the public is confused. Hopefully this
19 would be an accurate, neutral source of information, Department websites and brochures that
20 can be disseminated through doctors' offices and other venues I think would be helpful to the
21 public.

22 MS. BERRY: Hunt?

23 DR. WILLARD: I'm having a lot of difficulty seeing what this committee
24 can do, although in general I agree it's a problem and a priority. I mean, again, there are
25 academic organizations that have taken this on, there are government websites, both NIH and
26 DOD, et cetera, which are providing information for the public at large, and I come back to my
27 usual argument about genetic exceptionalism. I'm not sure that getting the public to weed out
28 the information that may be a little misleading or overstated in the area of genetics is not any
29 different, from my perspective, from what I see in cancer, weight loss, hair loss or hair gain
30 depending on your point of view, all the things we get on a daily basis in our email if we don't
31 have good filters.

32 So I'm having a hard time seeing why this is an issue for this committee as
33 opposed to simply acknowledging that it's an important issue and hoping that more people will
34 go to the good websites than they will to other websites.

35 MS. BERRY: Well, I was going to ask your exceptionalism question.
36 What is it about genetics that makes the public awareness issue that much more critical, or does
37 it? I mean, is there some unique aspect to genetics and genetic technologies that warrants some
38 sort of special federal or HHS role in public awareness that doesn't exist in other areas of
39 medicine?

40 DR. WILLARD: As you would predict, I don't see the issue that's specific
41 or unique here, but others I'm sure may.

42 MS. BERRY: David, then Ed.

43 DR. FEIGAL: There are two layers to think about, about public access to
44 your opinions. One is as experts on genetics, and that is very broad. But the other is the work
45 of the committee itself. This is a public committee, and although there is a small audience in

1 attendance at many public committee meetings, the ability to actually have an open and
2 transparent discussion of controversial issues and get them in front of the public is something
3 that's very unique in the United States. We're very aware of that at FDA because our advisory
4 committees are more heavily covered sometimes than our own decisions on the same products.
5 But it brings the debate to the public in a way that doesn't happen in Europe,
6 for example. In Europe, the decisions are just made, and even the basis for the decision can't be
7 reached by anything like FOI.

8 So part of my advice to you would be to use your bully pulpit. If there are
9 important messages about this area that need to get across, take advantage of the fact of the
10 stature of this committee, the stature of you as a group. Even though you're advisory to the
11 Secretary, it's in a public fashion. He didn't bring you in in a closed fashion. In fact, that was
12 done initially by using only government advisors. You have an opportunity to really leverage
13 what you want to do just through this kind of process.

14 MS. BERRY: Ed, and then Emily.

15 DR. McCABE: I was going to make three points. One is that in terms of
16 genetic exceptionalism, there are some things that we will take up in this committee that are
17 within the purview of our role dealing with genetics that aren't genetic exceptionalism, but we
18 are not advising the Secretary on all issues of health. So to the extent that they may be
19 particular to genetics and genomics, then that falls within our purview, and to single them out is
20 important. It doesn't make them exceptional for genetics.

21 Secondly, again to make the point, there's a lot of work that has been done
22 by SACGT that has not seen the light of day in terms of brochures and information that could
23 be useful to the Secretary, and completing that work, if we chose to do so, and bringing that
24 forth would be worthwhile.

25 Then the third point is that a lot of this discussion I really think is very
26 similar to the discussion about professional education and training, and that has to do with
27 being particularly sensitive to the diversity within our culture and making sure that not only 20
28 percent of the public is aware but as much of the public as we can possibly deal with can be
29 aware so that we deal with the issues of language diversity, cultural diversity, and try and be
30 sensitive to those needs as we educate the public.

31 MS. BERRY: I've got Emily, and then Francis.

32 DR. WINN-DEEN: I guess I wanted to address Hunt's comment. I think
33 the one place where genetics is different because of its nature is in its predictive nature. So if
34 you can say your genetic makeup means, even though you're symptom-free today, you're going
35 to have this disease tomorrow, and we do have some specific examples, like Huntington's,
36 where that is the case, I think then you're in a situation where people have to understand that
37 that's the situation and you don't want to be in a situation where people are testing for -- let's
38 just assume for the moment that this is not true, that you carry this gene, so therefore you're
39 obese and you'll always be that way and you can't do anything about your weight, so just resign
40 yourself to diabetes and heart disease and everything else.

41 Now, maybe there is a genetic component, but until that's proven and well
42 documented, we don't want people getting either the wrong health care or not getting the right
43 health care because they don't really understand what's going on and what's true and what's not
44 true. Now, a large part of that should be happening with a health care provider. But as you
45 mentioned, your email box is filled with misinformation, particularly around weight loss and

1 that area. I think if we're not careful about educating the public so they really at least know
2 where to go -- I mean, a public awareness campaign can be NIH has this website. It doesn't
3 have to mean that you do all the in-depth education in your primary format.

4 But I'm just concerned that if we don't do something, we don't make it a
5 priority that the end user ends up being uninformed.

6 MS. BERRY: Francis, and then Joan.

7 DR. COLLINS: I don't think anybody would disagree with the premise that
8 it is an important area, that public awareness of what genetics can do for you and what it can't
9 would be a good goal to try to achieve. I think the problem is it's very hard to figure out how to
10 do that in a fashion that gives people information when they didn't really think at the moment
11 they were looking for it anyway. So this notion of trying to identify the teachable moment, and
12 then be sure you have validated information available to the people who are at that moment and
13 looking for it seems like a fairly attractive strategy.

14 That, in fact, is one of the reasons that we have pushed, I think over the last
15 10 years, more in the direction of trying to educate the health care professionals, because many
16 people at the teachable moment go to the health care professional and say what should I do
17 about the fact that I just heard that my brother has been diagnosed with colon cancer? Does
18 that mean I should be getting screened? That kind of question.

19 We have, through NHGRI, and even more from the Department of Energy,
20 funded a fair number of public education projects through the ELSI program over the last 10
21 years, and they've been a diverse array of projects, some of which were simply public forums,
22 some of which generated materials, some of which supported PBS television shows that some
23 of you have probably seen.

24 The problem is it's really hard to evaluate what the impact of that
25 educational investment has been, and it may be that the investment has not yielded a big, huge
26 difference. Most people get their information about genetics either from the media in sort of a
27 fashion where they're filtering a lot of other things and may or may not be tracking this one or,
28 as I said, they seek it out at a time when they're specifically faced with a question about
29 themselves or their family and they're most likely then to ask their health care professional.

30 People who are looking for information on the web, you should certainly be
31 aware there's a lot of pretty good information about genetics and genomics that you can find on
32 a lot of websites. Probably the NIH website more than any other is loaded with information,
33 including some that is specifically designed for people from different cultures, or even from
34 different languages. It's not perfect. There are holes in it. It's not as broad and diverse as it
35 should be. But I think it is possible to get some information if you're looking for it.

36 I guess here, as some of the rest of you have said, while this is a very hard
37 problem, it's also a hard one to come up with an obvious mechanism to solve the problem,
38 particularly if we're not quite sure right now what the message is. I've always thought it was
39 useful to ask the question if somebody donated money to you, to have a 60-second spot in the
40 middle of the Oscars, just to be topical, about genetics, and you were the one to design that
41 particular 60-second public interest spot, what would you try to say to all of those people
42 gathered around their televisions? What would be the message you'd try to convey?

43 Genetic testing could help you? Well, that doesn't sound so good. Know
44 your family history. That would be a good thing for people to know about. Maybe that would
45 be a decent message, but it would be a little hard to get a lot of people to jump up off their

1 couch about that one. What is it that we want people to be aware of, and what actions do we
2 want them to take? I don't think we're even quite in consensus about the answer to that
3 question, which makes it hard to design a public awareness campaign.

4 It's much better, of course, if you had a circumstance where you really have
5 a message about AIDS prevention and you know what it is you're asking somebody to do.

6 MS. BERRY: Joan?

7 DR. REEDE: Well, the pediatrician in me is coming forward. There's a
8 part of educating the public at large, but there is for me a real part of how do we educate those
9 that are in our educational system, our K-12. I know that the briefing mentions it briefly and
10 says that there's a lot being done, but if you look at the standards, if you look at AAAS, if you
11 look at NSF, if you look at the state standards, the various standards, although it mentions
12 genetics, there is really no focus on human genetics. If you look at most of the classrooms, you
13 will spend an entire year studying all sorts of things and one day understanding anything that
14 relates to human genetics.

15 So I think there is a point to step in and say that understanding what goes on
16 in terms of human genetics is important. I think this dividing it up into standards being set by
17 NSF over here, NIH has developed some curricular pieces, but they are truly not integrated
18 across the board. There are a lot of school systems that don't know that they exist. The
19 agencies actually have to start working together. If a set of standards is being put in place over
20 in one area that completely ignores or minimizes the importance of human genetics, it is hard to
21 move this forward in terms of educating future generations about these issues.

22 MS. BERRY: Yes, Kimberly?

23 MS. ZELLMER: I just wanted to reiterate what Francis was saying. I think
24 that when we're talking about public awareness, I think there's a lot of information out there,
25 and I think that part of it is what the public is interested in. I think that personally, I'm an
26 educated person but I knew very little about genetics until my daughter was diagnosed, and that
27 obviously generated an interest, and I found plenty of information once I wanted the
28 information. I think it depends on what specifically you're trying to raise awareness with the
29 public, because I think if you're just talking sort of general genetic principles, those that are
30 interested have plenty of resources to go to and look for.

31 Those who don't really have an interest, I don't know how you're going to
32 get their attention. I don't know if you put a brochure in the doctor's office and they don't really
33 have any issues, I'm not really sure that that's going to generate any more public awareness. If
34 you provide forums for people who aren't aware of any genetic issues, I don't know how
35 effective that's going to be.

36 It seems like what we've talked about is sort of warning people about these
37 emails and false advertising and things like that, and I think that's kind of a different issue,
38 more than general public awareness. I mean, to me that seems more like how do we regulate
39 these advertisers and protect people from that than just sort of general public awareness,
40 because I think there's a lot of good resources if people want to know about genetics and
41 different types of genetic disorders.

42 But I think really what we're talking about is protecting people from the bad
43 information, and I don't know exactly how we would do that. But that seems more like the
44 advertising issue rather than necessarily public awareness.

45 MS. BERRY: Does anyone else have any comments? Yes, David, and then

1 Matthew.

2 DR. FEIGAL: Let me just give a quick counterexample. I think if you felt
3 strongly, for example, in the discussion we were having before lunch, that a large population
4 study was something that was unusual, it's going to take a lot of consensus building to get the
5 kind of trust for something like that to enroll rapidly. So to have an endorsement, to talk about
6 why that's a good idea, how that's going to make the investment in the Human Genome Project
7 pay off and so forth. There may be other people who say it, but don't underestimate the impact
8 that you can have, too.

9 MR. DAYNARD: I just wanted to add my suggestion that the committee
10 look upon public awareness and its input to that as an access issue. That's the way the Federal
11 Trade Commission looks at consumer awareness about any issue, particularly issues that affect
12 their health. If it's nothing more than telling consumers what the good websites are, or
13 examples of bad websites, it's going to affect their access, particularly when you get into
14 communities that may not otherwise have normal access. So that's the way I'd suggest looking
15 at it. Thanks.

16 MS. BERRY: Martha?

17 DR. TURNER: A quick process issue relative to what we're talking about
18 when we're talking about this, and that is that it's not so much that we know today what we
19 want the public to be aware of but that we agree that as we learn about things, the public needs
20 to become aware of them, if appropriate. So if we find something out that we should build into
21 whatever marketing plans and dissemination plans we have, some sort of piece for public
22 education that is not perhaps on the web, because there are a whole lot of people who don't use
23 computers today -- and so when we identify a population we want to know something, then we
24 need to develop ways to make sure that's a part built in, as opposed to something that may or
25 may not happen depending on the economics or the marketing.

26 MS. BERRY: Brad, did you have something?

27 MR. MARGUS: I'm not really sure how we can improve awareness. I like
28 the idea of using our pulpit as a committee, anything we decide to support like a large
29 population study, to use our opportunity to shout it from the mountaintops.

30 I just wanted to bring up one subject, though, and that is are there some
31 more things out there that we could endorse or that we could impress on the Secretary to talk to
32 other agencies about or to help support, such as at the last meeting when the FTC taught us
33 about how much can be enforced. Given the very, very restricted resources and budget, I
34 learned that people pretty much have to be dying from a claim, and even then it can't be on a
35 regional level, it has to be on a national level, all these things that made it really tough to
36 enforce anything.

37 That was pretty outrageous to me. So while I'm not exactly sure how we
38 can change awareness overnight in any big, broad stroke way, if there are certain things that we
39 could still emphasize so that when the Secretary is sitting in the Cabinet meetings this somehow
40 can be underscored, I think we need to speak up if it affects our area. In the case of the FTC
41 example, that really affected our area. I mean, here we are thinking about how can we educate
42 people better, but it turns out that even if people were out there making wild claims, from what
43 I understood from last time, it's pretty hard to enforce. Maybe we should just go on record as
44 saying that we're not happy with that.

45 DR. McCABE: We're going to need to wrap this up very quickly so that we

1 can move on to the public comment.

2 MS. BERRY: I was just going to wrap up by saying I think, based on the
3 issue briefs and our conversations, it seems like there is a pretty significant gap in public
4 awareness. There are public awareness efforts underway by different government agencies and
5 by private organizations. Are they sufficient or are there gaps? We don't really know, so the
6 question is while we think this is a very important issue, obviously because of the ranking that
7 it achieved -- it's fairly high up there -- what is our role? Without getting into specific
8 recommendations, are we going to talk about starting our own website? Are we going to have
9 forums? Whatever those may be, that's something for another day.

10 Do we want to have a more monitoring, passive role in keeping an eye on it,
11 declaring the priority that it is but sort of sitting back and watching these other activities? Or
12 do we want to have more of a leadership role and be more aggressive in using our bully pulpit
13 and coming up with concrete recommendations and partnering with different agencies and
14 groups? That sort of leads us to the discussion of the categorization. Where would we put this
15 in the 1 to 4 categories? I'd entertain anybody's motion for a category.

16 Kimberly?

17 MS. ZELLMER: I think this is one of the Category 3 is what I would say,
18 in that it transcends all issues and that on a case-by-case basis we should see it as to the issues
19 that we're discussing, whether it's something that maybe we want to make the public aware of,
20 or if there are aspects of whatever we're discussing, whether it's coverage and reimbursement or
21 large population studies or whatever, whether there is some public awareness aspect that we
22 want to make sure that the word gets out.

23 MS. BERRY: Yes, Barbara?

24 MS. HARRISON: I think I'd just add a thought that's been going through
25 my mind, which is really how pervasive and how important public awareness is. I mean, the
26 reason why we don't have underrepresented populations participating in studies is because
27 they're not aware of genetics. They don't know how important it is for them to be involved in
28 that clinical trial or for them to be involved in this research protocol, because it significantly
29 contributes to the development of drugs and that kind of thing.

30 So I think because we're here to serve the public and we want the public to
31 be aware of what we're doing, I think public awareness really has to be up there. I do struggle
32 with how is the best way to do it, but I definitely think that it's a priority issue and I can
33 definitely see it being put in 3, I guess myself.

34 MS. BERRY: Yes, Joan?

35 DR. REEDE: Along those same lines, I think one of the things that we
36 might consider as a committee, if we put something into a Category 2, which means we think
37 it's important enough that we need to act quickly or it needs to be monitored for a period of
38 time, or a Category 4, something that we need more information on, that we as a committee also
39 think about how do we make the public more aware of this issue. So as we send a letter to the
40 Secretary or use some other vehicle, at the same time think about the public. It may be that we
41 need to do more than have an open public forum meeting like this. Are there other things that
42 we need to do to inform people about the opinions that we have or the issues? To date, I don't
43 see us having really engaged in that part of the discussion as much.

44 DR. McCABE: Okay. Well, having heard the discussion -- thank you very
45 much, Cindy. Having heard the discussion, do I have a formal motion? It sounds to me like

1 this is moving toward a Category 3, transcends all issues, needs to be included in any
2 discussion of any issue about how to make the public aware of that. Can I have a formal
3 motion to that effect?

4 I'll take Kim, since you brought it up first, and Chris as a second to that.

5 Any further discussion of this?

6 (No response.)

7 DR. McCABE: All in favor, say aye.

8 (Chorus of ayes.)

9 DR. McCABE: Opposed, nay?

10 (No response.)

11 DR. McCABE: Abstain?

12 (No response.)

13 DR. McCABE: Okay. So that, then, is a Category 3, and with that, we'll
14 continue the discussion on these tomorrow.

15 But now it's time for us to hear from the public. This is something that's
16 extremely important for us. We've just been discussing that this is a public forum for
17 deliberations, and we value the input we receive from the public. We set aside a time during
18 each meeting for public commentary. There will be time today and tomorrow. There are also
19 written comments which appear under Tab 1 of your briefing book, and some additional have
20 been passed out today or are in your table folders.

21 I'll ask each of our public speakers to please limit your comments to five
22 minutes, and today we'll be hearing first from Dr. Margaret Gulley, Chair, Molecular Pathology
23 Committee, College of American Pathologists.

24 You can sit or stand, whichever you prefer.

25 DR. GULLEY: This is what the written comments look like, and there's
26 extra copies out in the hall.

27 Dr. McCabe and members of the committee, good afternoon. My name is
28 Margaret Gulley and I'm Director of Molecular Pathology in the McLendon Clinical
29 Laboratories at University of North Carolina Hospitals. I'm also a faculty member in the
30 Department of Pathology and Laboratory Medicine of the University of North Carolina at
31 Chapel Hill Medical School. Today I'm here as a representative of the College of American
32 Pathologists, or CAP, where I currently serve as Chair of the Molecular Pathology Resource
33 Committee.

34 The comments expressed by the CAP reflect a set of fundamental principles
35 regarding genetic testing and quality laboratory medicine. The purpose of these comments is to
36 provide the committee information with regard to genetic testing issues as they relate to the
37 priorities under consideration. Specifically, my comments will describe the College's
38 involvement in the area of CPT coding towards reimbursement for genetic technologies, the
39 CAP's position on patent policy impacting genetic tests and our progress on the College's
40 approach to address genetic test oversight utilizing existing regulatory mechanisms that include
41 laboratory accreditation and proficiency testing programs.

42 So first, on CPT coding on reimbursement, the College has formed a
43 Genetic Testing Work Group focused on creating appropriate code assignments for molecular
44 genetic testing for recommendation to the AMA CPT Editorial Panel. In an effort to better
45 utilize molecular genetic tests, the work group proposed to the AMA the implementation of a

1 coding system that will offer diagnostic granularity without changing test descriptions and thus
2 be less prone to payment denials.

3 The work group includes representatives from the American Society for
4 Clinical Pathology, the Association for Molecular Pathology, the American College of Medical
5 Genetics, the American Association for Clinical Chemistry, and the American Clinical
6 Laboratory Association. A consensus was reached that use of code modifiers in the current
7 CPT system would result in widespread acceptance from payers and enable providers to submit
8 specific information to adjudicate claims.

9 The College led a breakout session during the AMA's November CPT
10 advisory committee meeting to discuss this approach. The session included representatives
11 from the AMA CPT Editorial Panel, the Centers for Medicare and Medicaid Services, the
12 AAHP/HIAA, and Blue Cross/Blue Shield Association. After careful review of prior options,
13 the breakout session facilitators all agreed that the numeric alpha code modifier system is the
14 most viable solution. Further, the work group concluded that implementation of the modifier
15 option would provide an accurate method of reporting and identifying molecular genetic
16 testing, the ability to track utilization of genetic tests to capture diagnostic granularity, and
17 permit data tracking to determine the genes that are most often targeted for testing, the
18 frequency of the tests, as well as the types of genetic tests that are most utilized.

19 The work group confirmed that CPT modifiers would result in widespread
20 acceptance from payers. The work group's recommendations were presented to the CPT
21 Editorial Panel and subsequently to the CPT advisory committee members for comment, and
22 placed on the CPT Panel's February 2004 meeting agenda. The College cannot report on the
23 outcome of this proposal because the proposed CPT coding changes are confidential and
24 proprietary until they're finalized. Because code assignments and descriptions can change until
25 just before publication, the American Medical Association, which owns the CPT, asks that
26 participating organizations not publicly release detailed coding information until publication of
27 the CPT volume.

28 What can be stated is that after two years of intensive work to create a
29 numeric alpha modifier system for molecular genetic test coding, a major milestone was
30 reached this past month when the CPT Editorial Panel favorably considered the proposed
31 system. We're hopeful that the proposed changes will be published this October for the 2005
32 CPT coding book edition.

33 I will now turn to patent policy impacting genetic technologies. As medical
34 specialists in diagnosis of disease, the College recognizes that genetic testing is an area of
35 growth and change for pathology and for all of medical practice in the decades to come.
36 Pathologists therefore have a keen interest in ensuring that gene patents do not restrict the
37 ability of physicians to provide quality diagnostic services to the patients that they serve. The
38 CAP believes that gene patents pose a serious threat to medical advancement, medical
39 education and patient care.

40 When patents are granted, subsequent exclusive license agreements and
41 excessive licensing fees prevent researchers, physicians, and laboratories from providing
42 genetic-based diagnostic services. As a consequence, patient access to care is limited, quality
43 is jeopardized, and training of health care providers is restricted.

44 The field of molecular pathology uses genes and their mutations to predict
45 or diagnose disease. The list of diseases that can now be diagnosed or predicted from gene-

1 based tests is growing rapidly. Physicians and scientists can often easily translate the
2 fundamental information derived from studying the human genome into diagnostic genetic tests
3 and use these tests for patient care. Because information about gene sequences is so
4 fundamental to the understanding of specific diseases, patent holders can gain essential
5 ownership of diseases through patents. Exclusive or restrictive license agreements on gene-
6 based tests have been used to prevent physicians and clinical laboratories from performing
7 these tests as diagnostic medical procedures.

8 Patients suffer because diagnostic test services are less readily available and
9 affordable. Medical education and clinical research are also threatened. In fact, CAP members
10 have received cease and desist notification letters from patent holders indicating that continued
11 patient testing would be a patent infringement. Examples of diseases where testing has been
12 halted due to physicians receiving such a letter include breast cancer, Canavan's disease,
13 Charcot-Marie-Tooth disease, and Alzheimer's disease.

14 The recent trend of using patents to monopolize gene-based testing services
15 is a radical departure from historical precedent in clinical laboratories, and it works against the
16 goal of making these procedures widely accessible and affordable to the public.

17 DR. McCABE: Could you wrap it up fairly soon, please?

18 DR. GULLEY: Sure.

19 Especially troubling is the fact that under patent protection, the
20 understanding of the utility of the test, as well as the underlying disease processes, also become
21 proprietary, thereby imposing a profound change in how the profession and the public acquire
22 knowledge about these tests.

23 In 1996, Congress recognized that medical procedure patents might impede
24 the advancement of medicine, curtail academic access, and place unreasonable limits on the
25 research community, and interfere with medical education and the quality of care provided to
26 patients. As a result, in October of '96 legislation was signed into law, the Frist-Ganske
27 amendment, that permanently precludes the filing of infringement suits against physicians and
28 other medical practitioners for the performance of medical activities that would otherwise
29 violate patents on medical or surgical procedures. A medical activity is broadly defined to
30 include the performance of a medical or surgical procedure on a human body, organ, or
31 cadaver, or on an animal used for research.

32 However, the act does not explicitly affect enforcement of biotechnology
33 patents or extend to clinical laboratory services. With the advent of new and innovative
34 approaches to gene-based diagnostic testing and the promise of enhanced and expanded
35 diagnostic testing, laboratory services and clinicians should have the same protection from
36 patent infringement as other medical providers and other procedures.

37 We're facing the unprecedented situation in which a single patent --

38 DR. McCABE: I'm sorry, but we have your testimony in written form. If
39 you could just move to the conclusion quickly, please.

40 DR. GULLEY: Okay. We believe that patents set an extraordinary and
41 dangerous precedent, and they affect the availability of diagnostic testing.

42 The final thing I wanted to touch on was genetic testing oversight. We have
43 presented an approach to the oversight of genetic testing that builds on the existing clinical
44 laboratory improvement amendment, or CLIA, laboratory inspection and accreditation process
45 to provide oversight and approval of genetic testing in lieu of federal regulations.

1 So we would like to suggest that instead of increasing federal regulations or
2 developing reduplicative federal programs, that we work through the existing programs to
3 improve oversight and federal oversight of genetic testing.

4 So you have the rest of my comments in the handout, and I would like to
5 conclude with saying that the College of American Pathologists appreciates the opportunity,
6 and we're here to answer questions or work with you as you continue to work on policy issues.

7 Thank you.

8 DR. McCABE: Thank you very much.

9 I think in the interest of time we need to move on to the next presenter.

10 That is Dr. Judith Lewis, who is president of the International Society for Nurses in Genetics,
11 or ISONG, and also a professor in the School of Nursing at Virginia Commonwealth
12 University, and a member of the Secretary's Advisory Committee on Genetic Testing.

13 Welcome back, Judy.

14 DR. LEWIS: Thank you, Dr. McCabe. I must say it's really nice to see that
15 some of the work that we did that didn't get finished is also being considered by the current
16 committee, so I thank you for that.

17 But I'm here this afternoon as the current president of ISONG, the
18 International Society of Nurses in Genetics. Our membership spans six continents and includes
19 nurse clinicians, nurse educators, and nurse researchers. ISONG is a specialty nursing
20 organization dedicated to caring for people's genetic health through excellence in the provision
21 of genetic health care services by fostering the professional and personal growth of nurses in
22 human genetics.

23 There are over 2.7 million nurses in the United States. Of those,
24 approximately 2.2 million currently are practicing as registered nurses. Approximately 7.3
25 percent of those, or slightly fewer than 200,000, are advanced practice nurses. Half of these, or
26 about 100,000, are nurse practitioners who are delivering primary health care services.
27 Compared to other primary health care providers, nurse practitioners are more likely to be
28 practicing in sites serving patients who are economically or socially disadvantaged or in
29 medically underserved areas. The average salary for a nurse practitioner in the United States is
30 slightly over \$60,000.

31 Of the 2.2 million practicing nurses, over two-thirds of us work at inpatient
32 hospital settings, with the vast majority of those involved in direct patient care. In the inpatient
33 setting, the nurse is the health care professional who spends the largest amount of time in direct
34 contact with the patient. The nurse is often the health care professional who first notes the
35 dysmorphic features of the newborn, who provides the patient with education about the nature
36 of a newly-diagnosed chronic condition, who answers the patient's questions about the meaning
37 of this illness for themselves and their family members, and who deals with the entire spectrum
38 of the human response to health and illness.

39 The nursing workforce holds great potential in caring for people's genetic
40 health. ISONG has, in conjunction with the American Nurses Association, developed and
41 promulgated the Scope and Standards of genetics clinical nursing practice. This document,
42 which is currently being revised and expanded, delineates the genetic competencies for nurses
43 practicing at the basic level, as well as enhanced competencies for advanced practice nurses. In
44 addition, the Genetic Credentialing Commission, an affiliate of ISONG, offers the advanced
45 practice nurse in genetics credential to Master's prepared nurses in specialty genetic services,

1 and the genetics clinical nurse credential to the baccalaureate prepared nurse who managed
2 genetic information in a variety of health care settings. These credentials are awarded on the
3 basis of a professional portfolio.

4 ISONG is committed to working towards ensuring that the nursing
5 workforce is well prepared to serve the patient's and the public's need for genetic information.
6 There are several programs designed to prepare nursing faculty, who may have been educated
7 themselves in the pre-genomic era, with the knowledge and skills that they need to include
8 genetics content in undergraduate and specialty nursing curricula. The National Institute for
9 Nursing Research offers a highly competitive, intensive fellowship designed for doctorally
10 prepared nurses, doctoral students, and advanced practice nurses. The goal of this summer
11 institute is to prepare nurses to become clinicians and researchers in the area of genetics.

12 Current programs, while providing a valuable service, do not have the
13 capacity to meet the demand. If we are to continue to prepare an educated workforce, the
14 profession needs resources to enhance its education and outreach efforts.

15 ISONG is committed to ensuring that all individuals have appropriate
16 access to genetics and genomic health care and has approved a position statement defining the
17 role of the nurse in ensuring access. I have provided you all with copies of this statement that
18 just was published this week. In addition, I have with me a single copy of the Scope and
19 Standards document for your use.

20 ISONG is eager to work with the Secretary's Committee on Genetics,
21 Health, and Society as you define your priorities and begin your work. We look forward to
22 providing you with information on the genetics nursing workforce, the way genetics nurses
23 practice in the United States and throughout the world, and with knowledge of the resources
24 that will be required to ensure that our over 2 million nurse colleagues have the knowledge and
25 skills they will need to practice effectively.

26 Thank you.

27 DR. McCABE: Thank you.

28 We have time for one or two brief questions, if there are any questions or
29 comments.

30 (No response.)

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

32 Our next speaker is Sharon Terry, president of the Genetic Alliance.

33 MS. TERRY: My name is Sharon Terry, and I'm president and CEO of the
34 Genetic Alliance. I want to thank you for this opportunity to offer comment and for your work
35 on these issues.

36 You've identified 12 important issues. Some, as you have noted, are subsets
37 of others. Prioritizing them can only be done in the context of a framework. By what metric
38 should these issues be measured and weighted? I propose articulating the metric, and it is
39 improved human health.

40 There are some premises by which we in the genetics community operate.
41 First, genetics is significant for more than just the "gee whiz" factor. It has something to do
42 with human health. Second, success for the science of genetics means translating this body of
43 basic knowledge into technologies and treatments that improve human health. Third, the
44 government should be involved in facilitating success. Fourth, genetics has engendered these
45 discussions, which set it apart from other basic science to health translations. So whether

1 genetic exceptionalism is right or wrong, it exists. Against this background, I'd like to
2 comment on these issues.

3 But first, a disclosure. I have a huge conflict of interest and an overarching
4 agenda. I speak mindful of the millions of individuals affected by genetic conditions. I know
5 what I know because I, as one among them, have worked alongside them for over 10 years. I
6 know what I know because my colleagues, other lay advocacy group leaders, face the loss of
7 their child, face the enormous impact of disabilities, face the inadequacies of the health care
8 system. I know what I know because my two children face blindness and a host of other
9 difficulties as a result of causal mutations in a gene.

10 I live with the issues you have laid out. I have discussed them in numerous
11 federal advisory committees, analyzed them over drinks, written papers about them. I think that
12 after the collective work of this community and all of your work, I think that the issues you
13 define are only symptoms, symptoms of a disease that needs to be described, and we, like so
14 much of medicine, are much more comfortable dealing with the phenotype rather than the
15 etiology of the disease. I believe that it is the challenge of this committee, if you want to make
16 a difference, to uncover the basic roadblocks and not continue to just describe the symptoms.

17 In fact, as several people have noted both here and in written comments,
18 many of the issues are a subset of access. Coverage and reimbursement, genetic discrimination,
19 genetics education and training, oversight, direct-to-consumer advertising, patents, and public
20 awareness are all subsets of access. Many of these issues are examined in a kind of isolation
21 that does not reveal the underlying cause, and many of them are examined in a political agenda-
22 driven light. I don't think this committee or any federal advisory committee has the resources to
23 recommend solutions to these problems.

24 Two of the other issues are a step closer to considering the major priority.
25 If the reason we care about genetics is because it will lead to improved human health -- I've
26 already disclosed that I have an agenda -- then we must put the symptoms together for a
27 diagnosis. The questions that pharmacogenomics and large population studies raise are related
28 and are closer to the root of the problem. Neither can be done well in the current regulatory
29 climate. Both are impeded by important protections that are misguided in implementation, thus
30 thwarting the very research we need to move an enormous body of basic science towards
31 translational research.

32 The steps along the way, meaningful epidemiology, natural history studies,
33 longitudinal studies, environmental studies, gene/environment studies, are all thwarted,
34 cumbersome, and de incentivized. This committee has a bully pulpit that can have an impact on
35 policy recommendations that could facilitate the climate necessary for these studies.

36 The ultimate questions are ones of integration. How will genetics be
37 integrated into medicine? How will scientific evidence be integrated into policymaking, payer
38 decisionmaking, agenda-setting for research priorities? Right now, the system in place has no
39 incentive for physicians to be early adopters of proven genomics technologies, for payers to pay
40 for new technologies and treatments, for researchers to strive for health outcomes as the
41 endpoint, for industry to take risks that will benefit marginalized communities, be they racial,
42 ethnic, or rare disease communities.

43 So the answer to genetic exceptionalism is an easy one. Genetics should be
44 integrated, and the path to integration probably involves both segregation and affirmative
45 action. The question before this committee is whether you are ready to be bold, to look at these

1 issues without the lens through which you normally look. Are you willing to go beyond the
2 symptoms to understand the etiology of the disease? Are you committed to discovering the real
3 roadblocks in the system that creates all these other issues and grapple with the system, not just
4 the symptoms?

5 This leaves you with the issue of a vision statement, an issue which could
6 be considered without substance. I contend that if you cast aside your usual imaging tools and
7 look at the whole patient in the context of a community, you have the brain power on this
8 committee to formulate a vision of genetics integrated, of a pathway to translation, of a future
9 where genetics and genomics improve human health. You have the ability to recommend
10 systems whereby politics no longer set the scientific agenda and basic science no longer holds
11 policy hostage.

12 I strongly suspect the answer might include universal health care, and while
13 you may feel this is beyond the scope of the committee, I suggest that not to name the disease
14 increases morbidity. We, the people who live with genetic conditions every day, who watch
15 our children die, who care for our sick siblings and parents, and who are limited by disease
16 ourselves, know well what the rest of the world will come to know, that science will never step
17 up to the plate and set a health outcomes agenda on its own, and that politics will never
18 understand the complexity of the system without the evidence that science offers.

19 It is time for the two to be integrated, to formulate, based on hard evidence
20 from all the sciences, a vision for the future. You must identify the roadblocks and recommend
21 the treatments. It is time for us to engage in the future for which we hope. We are ready. We
22 hope you are, too.

23 Thank you for your service, your thoughtfulness, and your dedication to
24 genetics, health and society.

25 DR. McCABE: Thank you.

26 I think, unfortunately, it's necessary for us to move on.

27 Our last speaker is Dr. Andrea Ferreira-Gonzalez, who is director of the
28 Molecular Diagnostics Lab and Associate Professor of Pathology, Virginia Commonwealth
29 University.

30 DR. FERREIRA-GONZALEZ: Dr. McCabe, members of the committee,
31 good afternoon. My name is Andrea Ferreira-Gonzalez. I'm a director of the Molecular
32 Diagnostics Laboratory and Associate Professor of Pathology at Virginia Commonwealth
33 University. I'm currently past chair of the professional relations committee for the Association
34 for Molecular Pathology, and I speak to you today as a representative of AMP.

35 The purpose of these comments is to provide information to the committee
36 on issues that are affecting the ability of laboratories to provide genetic testing services. There
37 are three major issues that I will address today. First is the inadequacy of the CPT coding and
38 reimbursement for genetic tests. Second is the negative effect of gene patents on molecular
39 diagnostic laboratories. And the last issue is an update on advances in the oversight of genetic
40 testing laboratories.

41 With regards to the first issue, CPT coding and reimbursement for genetic
42 tests, a widely held view which AMP upholds is that molecular genetic tests will influence all
43 aspects of medical practice in the future as we unravel the variations in the human genome that
44 correlate with disease and disease risk. Molecular genetic testing must be financially viable so
45 that physicians and patients may realize all the diagnostic benefits of our understanding of the

1 human genome.

2 Currently this is not the case, and several factors prevent the appropriate
3 cost recovery for genetic testing laboratories. These factors are the inadequacy of the CPT
4 coding system available for the billing of molecular genetic tests and the low reimbursement
5 levels set for the CPT codes that are available and currently in use. The Association for
6 Molecular Pathology has been working closely with a Genetic Testing Work Group over the
7 last two years, with the goal of developing and implementing an appropriate billing code
8 system for molecular genetic tests.

9 The College of American Pathology chairs the work group with members of
10 other associations. The work group has proposed to the AMA implementing a coding system
11 that will provide payers with more specific information about genetic tests performed, and thus
12 be less prone to denials. The genetic working group's recommendations were presented to the
13 CPT Editorial Panel and subsequently given to the CPT Advisory Committee members for
14 comment. The proposal was discussed at the CPT Panel's 2004 meeting, and AMP understands
15 that the recommendations were received favorably.

16 AMP asks that the SACGHS remains cognizant of the progress in
17 implementing the proposed changes in molecular genetic test coding and the impact on
18 payment for molecular genetic testing in the future.

19 The second factor affecting the financial viability of molecular genetic
20 laboratories is the current Medicare reimbursement levels set for existing CPT codes. These
21 reimbursement levels, which will be presented later on today, are far less than the cost of
22 performing molecular genetic tests. The reimbursement levels were set more than 10 years ago
23 and were inadequate even then. They do not reflect the current cost of genetic tests, they are
24 technically complex and frequently require highly detailed analysis and interpretation. So
25 AMP asks SACGHS to consider steps that could provide reasonable cost recovery for genetic
26 tests.

27 The effect of gene patents on genetic testing services. The granting,
28 licensing and enforcement of gene patents is having a broad negative impact on the ability of
29 clinical laboratories to perform genetic tests. While licensing fees may be financially
30 devastating to a molecular genetic laboratory already facing inadequate cost recovery for
31 genetic tests, even more egregious is exclusive licensing or enforcement resulting in a sole
32 provider of a medical service.

33 AMP strongly holds that a sole provider of a medical service is not in the
34 best interest of the public health. Examples of diseases where testing has been halted due to
35 patent or licensing enforcement includes breast cancer, Alzheimer's disease, Canavan disease,
36 and Charcot-Marie-Tooth disease. The growing trend of using patents to monopolize genetic
37 testing services severely compromises the accessibility and affordability of genetic tests for the
38 public. Particularly troubling is that under patent protection, the growth in understanding of the
39 utility of a genetic test, and even the underlying disease processes, also become proprietary.

40 Congress already recognized that medical procedure patents impede the
41 advancement of medicine, curtail medical access, places unreasonable limits on the research
42 community, and interferes with medical education and the quality of care provided to patients.
43 As a result, in October 1996, legislation was signed into law that permanently precludes the
44 filing of infringement suits against physicians and other medical practitioners for the
45 performance of medical activities that would otherwise violate patents on medical and surgical

1 procedures. However, the law does not cover biotechnology patents and does not extend to
2 clinical laboratory services.

3 In 2003, Representative Lynn Rivers, who was not re-elected, introduced
4 legislation that would protect physicians and other providers of clinical laboratory services
5 against enforcements of gene patents and against liability for infringement of patents on genes.
6 The logic of the bill was that genetic test services are part of medical practice and should be
7 widely available to promote optimal patient care, medical training, and medical research.

8 We anticipate that a new bill will be introduced in Congress to address this
9 concern. AMP encourages the Secretary's committee to examine the negative impact on
10 medicine of current practices in the patenting and licensing of genetic sequencing and work to
11 eliminate restrictions on the medical use of genetic information.

12 The last topic I would like to point out is genetic testing oversight. The
13 Association for Molecular Pathology has worked closely with the Food and Drug
14 Administration's in vitro diagnostic division to provide a laboratory perspective on the previous
15 committee's proposal for FDA oversight of laboratory-developed genetic tests. AMP supports
16 the existing clinical laboratory improvement amendment, laboratory inspection and
17 accreditation process to provide oversight and approval of genetic tests in lieu of new federal
18 regulations, and even FDA oversight.

19 A major concern of the previous committee in the area of oversight was the
20 review of laboratory-developed tests prior to their coming into clinical use. In response to
21 these concerns, the College of American Pathologists has developed and added test validation
22 questions to the molecular pathology checklist that will be used for inspections in the future.

23 AMP asks the Secretary's committee to review the changes implemented by
24 the College of American Pathology addressing test validation oversight concerns to determine
25 if these changes address the concerns raised by your predecessor.

26 The laboratory inspection process is only as good as the inspectors
27 performing the reviews of the laboratories. AMP has hosted training sessions for molecular
28 pathology laboratory inspectors at its previous three annual meetings, since AMP members are
29 among the experts in molecular testing. AMP supports the training and use of qualifying
30 inspectors to strengthen the review provided to molecular laboratories during the inspection and
31 accreditation process.

32 Furthermore, AMP's clinical practice committee is addressing clinical
33 practice issues such as developing a consensus on information to be included in a test report for
34 common genetic disease.

35 On behalf of AMP, I thank you for the opportunity to speak to you today.
36 AMP remains available to you to assist you with or provide information for your thoughtful
37 deliberations and important work.

38 Thank you.

39 DR. McCABE: Thank you.

40 I think we need to move on because of the pressure of time. We thank all of
41 our members of the public for presenting to us today. We very much appreciate your input.

42 Now we're going to begin a series of presentations on coverage and
43 reimbursement of genetic technologies and services, and that will be followed by a roundtable
44 discussion. I'd like to invite the presenters for the afternoon session to please join us at the
45 table. Your names are there at your places.

1 DR. LEONARD: Ed?

2 DR. McCABE: Yes?

3 DR. LEONARD: Can I ask a question? Can we get a copy of the
4 comments from the woman from Genetic Alliance, please?

5 DR. McCABE: Sharon, had you provided us with your comments?

6 MS. TERRY: I had to do them on the airplane coming here, so I will email
7 them to Sarah. Is that okay?

8 DR. McCABE: Okay. So the comment, just for the record, is that they will
9 be made available to Sarah as soon as possible. Thank you very much. I think they are
10 important.

11 Do you have them on a disk?

12 MS. TERRY: I have them on my laptop. I can bring them to the business
13 session.

14 DR. McCABE: Perhaps you could work with one of the staff members to
15 get them transferred over so we could have them printed out, because I think they will inform
16 further discussion tomorrow.

17 I think, as all of you will recall, the committee really decided that this was
18 an important issue, coverage and reimbursement, at our October meeting, and will help add
19 background to our priority-setting deliberations, which will continue tomorrow morning. I
20 want to thank all of the presenters for taking time from your busy lives to be here with us. We
21 look forward to an enlightening series of talks that are going to come at this from a variety of
22 different perspectives.

23 Because of the time constraints, I'm not going to go through extensive bios.
24 Those are under Tab 1 in your briefing books, and copies of the presenters' PowerPoint slides
25 are in the table folders, this white folder that was at your position this morning.

26 So our first presentation of the afternoon is by Dr. Michele Schoonmaker,
27 who will brief us about coverage and payment decisions and how they're made for genetic
28 technologies and services by private health plans. Dr. Schoonmaker is on the staff of the
29 Congressional Research Service of the Library of Congress. This group provides objective,
30 bipartisan analyses to Congress on genetics issues.

31 Dr. Schoonmaker?

32 DR. SCHOONMAKER: Good afternoon, Mr. Chairman, members of the
33 committee, and the public. Thank you for inviting me to participate in this session on the
34 coverage of genetic tests and services. The views I will present are my own and do not reflect
35 the views of the Congressional Research Service or the Library of Congress.

36 I'll begin by briefly describing health insurance coverage in the U.S., and I'll
37 focus mainly on how private insurers make decisions to cover new genetic tests. Then we'll
38 provide a general overview for how they pay for them. Throughout, I will give examples of
39 existing coverage policies for genetic technologies.

40 In 2002, almost 44 million people were uninsured. Seventy-two million
41 were covered by a public program, such as Medicare or Medicaid or the military, while the
42 majority, or 199 million people, had private insurance. Of those with private insurance, 88
43 percent were covered by group policies, usually through their employers, and 12 percent
44 purchased individual policies. The percents don't add up to 100 percent because there are about
45 29 million people who were counted in more than one group during the year, for example that

1 moved between being uninsured and being on Medicaid.

2 There are two main types of private insurance products. In indemnity
3 insurance, the insurer provides financing only. The insurer pays a provider a fee for service
4 after the patient has received the service. Because indemnity insurance originated to protect
5 people from the high cost of injury or illness, historically benefits excluded preventive services.
6 Most of today's indemnity plans have adopted some element of managed care.

7 Managed care emerged in the 1980s as an incentive to reduce the cost of
8 health care. Managed care insurers do this by coordinating the financing of care with the
9 delivery of services. Prior authorization and case management are two of the tools used to
10 encourage patients to see certain providers or to control their access to certain types of services.
11 For prior authorization, a patient has to get payer approval before seeking care. In case
12 management, a provider coordinates care through referrals. These practices aim at ensuring
13 that a patient receives only medically necessary services. Unlike indemnity insurance, most
14 managed care products generally include some coverage for preventive services.

15 Managed care products span a continuum of possible arrangements. Health
16 maintenance organizations are the most managed in that they completely integrate the financing
17 and delivery of services. Physicians are usually salaried employees or paid per member per
18 month regardless of whether or not a patient actually seeks care. HMO patients must see HMO
19 network providers or care generally won't be covered. In a provider organization, the payer
20 contracts with networks of physicians for primary care specialty services. The provider and the
21 payer negotiate discounted fees in exchange for a higher volume of referrals.

22 A point of service plan is a hybrid between the HMO and the PPO
23 arrangement. The point of service evolved to give patients more freedom in their choice of
24 provider. The patient can choose an in-network provider and have HMO-like benefits with low
25 co-payments, or they can choose to go out of network but with higher co-payments.

26 I've included the last two slides because it's important to understand the
27 distinction, because the type of organization can impact how coverage decisions are made and
28 how the providers are reimbursed.

29 I'll now discuss how coverage policies are made and provide examples of
30 existing policies. To prepare for the upcoming slides, I looked at the websites of 125 private
31 health insurers. Though 44 of them posted their coverage policies, only 27 allowed unrestricted
32 access. Of the 27, 24 had posted policies related to genetic tests or services. Sixteen of these
33 were Blue Cross/Blue Shield plans, and eight were other companies.

34 Insurers make coverage decisions in two main ways. The insurance
35 contract or a policy that an individual or employer can purchase can outline a broad benefit
36 category, such as laboratory tests. Decisions about a specific test are then made on a case-by-
37 case basis when the claims are processed. Alternatively, insurers can develop a coverage
38 policy. The policy is a more precise description of the exact service that will be covered, and
39 the conditions for which it will be covered. Policies are usually developed to respond to new
40 technologies, to new information about a technology, or in response to mandates. Policies
41 define what the insurer considers to be medically necessary, and they state limits on the types
42 of providers that can perform the service or can limit the number of times a patient can receive
43 a service. The coverage policy is written as a guideline. The exact benefits for an individual
44 are usually still determined by what their insurance contract says.

45 For almost all private insurers, a medical director will decide the benefits,

1 sometimes in conjunction with a medical policy advisory committee. The committees also
2 often include other plan personnel, as well as local medical experts, consumers, or legal
3 counsel. Employers can decide benefits. Regardless of what an insurance coverage policy
4 says, the employers, usually the CEO or human resources director, can negotiate with the
5 insurer for inclusion or exclusion of specific services. Other large groups, such as unions,
6 churches, academic centers, can negotiate benefits on behalf of individuals; and, of course,
7 federal and state governments can mandate coverage.

8 What criteria do plans use to determine what services to cover? Almost
9 unanimously, decisions are based on medical necessity. However, like beauty, medical
10 necessity can be in the eye of the beholder. While most plans don't publish specific definitions,
11 others have developed very explicit criteria. The following are the Blue Cross/Blue Shield
12 Association's criteria for deciding coverage.

13 First, the technology must have final approval from the appropriate
14 regulatory body. Once approved by the body, the TEC, or technology evaluation center, is not
15 bound by the indications in the approval. They can evaluate off-label uses.

16 Second, the scientific evidence must permit conclusions concerning the
17 effect of the technology on health outcomes. The evidence is evaluated in terms of quality and
18 consistency of results. The evidence should demonstrate that the technology can measure
19 changes related to the disease, and that the measurements actually affect the outcomes. The
20 technology must improve the outcome, and the benefit must be as big or bigger than established
21 alternatives, and be attainable outside of an investigational setting.

22 So in order for Blue Cross/Blue Shield to recommend coverage for a new
23 genetic test, the test must have FDA approval or conform to the CLIA requirements, evidence
24 must show that the test measures what it's supposed to measure, and that the test will positively
25 impact patient outcomes in the real world.

26 I want to point out that criteria 3 and 4 go beyond what is necessary for
27 regulatory approval of a new test. Rarely would FDA require that an applicant measure actual
28 clinical outcomes, let alone determine the magnitude of benefit in order to gain approval.
29 CLIA regulates the process of testing but doesn't say much about the evaluation of the test
30 itself. And although Blue Cross/Blue Shield doesn't explicitly use cost-effectiveness, one or
31 two other insurers considered cost-effectiveness but they didn't describe how the criterion is
32 applied.

33 So where do payers look for scientific evidence? Looking at the reference
34 section of the existing coverage policies, payers primarily rely on the literature, statements from
35 professional organizations, and government agencies. Many turn to the work of technology
36 assessment groups, and some payers adopt all or part of the policies from other payers.
37 Compared to work I did only three years ago, many more payers are turning to the websites as a
38 primary source of information.

39 Looking at existing policies, private insurers generally find genetic testing
40 medically necessary when personal or family history indicates a high risk for inherited
41 conditions, when the sensitivity of a test is known, when the results will directly impact the
42 treatment or management of the patient, when the diagnosis remains uncertain following
43 conventional workup, and interestingly, where pre- and post-test counseling is provided as
44 appropriate. A few policies even went so far as to specify what the informed consent should
45 be.

1 In general, genetic testing was not covered for population screening without
2 a personal or family history, regardless of ethnicity. The only notable exception was coverage
3 for CF carrier screening as a preconception service to what one policy called "informed
4 couples," or as a prenatal service to pregnant women. Testing is not covered for informational
5 purposes only or for testing minors for adult-onset diseases. Coverage is usually not provided
6 for a patient's family member who is not also a member of the health plan unless the
7 information from that family member, such as the identification of a specific mutation, is
8 necessary to make an appropriate medical decision for the plan member, and the family's
9 member can prove that they've already tried to get insurance coverage from their insurance
10 company and they were denied.

11 Nearly all insurers cover tests for chromosomal abnormalities. Most are
12 written for prenatal or neonatal diagnosis, but some insurers have also written policies for
13 preimplantation diagnosis. Covered indications include advanced maternal age, suspected fetal
14 anomaly, history of multiple miscarriage or developmental problems, et cetera. Tests for rare
15 single-gene disorders are usually covered under general policies for genetic testing and
16 counseling. However, some insurers have separate policies for specific conditions, and some of
17 those are for hereditary cancer testing, cystic fibrosis, Tay-Sachs, or hemochromatosis.

18 Policies for pharmacogenetic or pharmacogenomic tests can be written
19 either in the context of the drug policy, such as for Herceptin, or they can be written separately,
20 one policy for the drug, another policy for the test. Some drug policies simply list the
21 prescribing characteristics, such as HER-2-positive, without saying how the characteristic is to
22 be determined.

23 I'm going to give you a few examples of policies that have been written.
24 You might be surprised to find out that 12 insurers have written a policy for testing for the
25 genetic markers associated with familial Alzheimer's disease, but none of the policies cover the
26 test. Using primarily the Blue Cross/Blue Shield criteria, these insurers concluded that testing
27 is investigational. There is insufficient information to demonstrate that the genotypes are
28 associated with Alzheimer's disease with a high positive predictive value.

29 The second example concerns colon cancer testing. There are two kinds of
30 hereditary colon cancer. Hereditary non-polyposis colon cancer is typically diagnosed based on
31 family history and is associated with mutations in two main genes, the MLH1 and MSH2.
32 Familial adenomatous polyposis, or FAP, is based on personal signs such as the presence of at
33 least 20 polyps in the colon. The patient usually has at least one first-degree relative with the
34 disease. FAP is associated with mutations in the APC gene.

35 Sixteen insurers have developed policies for genetic testing. Four cover
36 testing without specifying the genes. Five cover mutation analysis in three common genes, four
37 cover microsatellite instability analysis in addition to the common gene tests, one covered APC
38 testing only, and two did not cover genetic testing but would cover other means of diagnosis.
39 Common exclusions in the policies were microsatellite instability analysis in the stool
40 specimen, and specifically testing for the I1307K mutation in the APC gene.

41 The point with this example is that even though 14 out of the 16 insurers
42 that had policies covered the genetic testing, they varied in the level of detail and also in the
43 specific analyses that they covered.

44 To give you a pharmacogenomic example, azathioprine is an
45 immunosuppressant treatment for inflammatory bowel disease. It is converted into active

1 metabolites by an enzyme called TPMT. The activity of TPMT is associated with genotype.
2 Ninety percent of patients are homozygous for wild-type form of the enzyme and have high
3 TPMT activity. Ten percent are heterozygous for a mutation that reduces enzyme activity, and
4 overall they have an intermediate activity. Finally, a small percentage are homozygous for the
5 mutant phenotype, and these patients have extremely low enzyme activity and are at risk for
6 toxicity.

7 People who are homozygous wild type could receive a standard dose of the
8 drug, and although no studies have measured the outcomes based on knowing the genotype
9 beforehand, the thinking is that a provider would prescribe a different drug or monitor
10 metabolite levels for those with the homozygous mutations, or reduce the dose for
11 heterozygotes. Six insurers have written policies for genotyping the TPMT gene. Three
12 covered both genetic testing and monitoring of the metabolite markers. Two covered only the
13 metabolite markers, and one did not cover either test.

14 Once an insurer decides to cover a new test or service, they have to
15 determine how much they're going to pay for it. Payment rates are based on many factors,
16 including where the services are provided and the usual and customary charges billed by the
17 providers in that location. Reimbursement rates can be a simple percentage of bill charges that
18 is determined in the insurance contract, insurers can negotiate fees with different providers, or
19 adopt a fee schedule such as those used by Medicare, or they can come up with their own fee
20 schedule. Just because a policy said that a test is covered doesn't mean that it will be paid at a
21 rate the providers find adequate. The reality of the situation is that poor payment can have the
22 same impact as a non-coverage decision.

23 The payment process involves recognition of a service by the insurer. This
24 is done through the common coding systems. The ICD-9 code tells the payer why a service was
25 done. It codes for the diagnosis, disease condition, signs or symptoms that the patient has. The
26 CPT identifies the procedure or the service that was performed. Basically, if the CPT, or the
27 service, matches the medically appropriate reason, or the ICD-9, then the claim can be paid. Of
28 course, this depends on the provider submitting first the correct information, especially with
29 regard to identifiers and dates of service, and second, where it's required, documentation. The
30 documentation could be a pedigree to show family history, or literature to support the medical
31 necessity of a new service.

32 The CPT codes used to identify genetic tests are limited. There are codes
33 for molecular diagnostic procedures and for cytogenetics. Usually you have to string together
34 multiple codes to describe a whole test. Providers often complain that these unbundled codes
35 have lower payments associated with them than would a code for a whole test. There have
36 been problems in the past as to what laboratory specialties can use which codes, particularly the
37 molecular codes that are in the cytogenetic section.

38 Each coding section ends with a code that's three digits plus 99. The 99
39 codes are for unlisted or new procedures. These are often perceived by insurers to be
40 investigational, indicating that we need timely development of new codes. As many in the
41 audience know, getting a new code can take years. The HCPCS, or Health Care Common
42 Procedure Coding System, is a national temporary coding system that can be established for
43 new tests. Many have recently been developed for gene sequencing and mutation analysis for
44 specific conditions. For example, you'd use an S3820 for a complete BRCA1/BRCA2 gene
45 sequence analysis, and an S3822 for a single mutation analysis for an individual with a known

1 BRCA1 or BRCA2 mutation in their family. Later, my colleagues will go into more detail
2 about payment issues with laboratory tests and with professional services.

3 So with that, to sum up, testing for the most traditional inherited genetic
4 conditions is covered by most private insurers, including counseling, but that doesn't mean that
5 there's adequate reimbursement. There still seems to be the perception that insurers are slow in
6 covering new technologies. Insurers may argue that this is because there isn't any data to
7 support the medical benefit of the new test. This situation may in part be due to the fact that
8 the studies designed to meet regulatory requirements rarely evaluate whether the information
9 from the test will impact patient management, and if so, how.

10 As far as payment is concerned, providers need to go through the cost
11 analysis with insurers so that payers understand how costs are applied and where
12 reimbursement rates are failing. Like many other things, at the heart of the issue is a need for
13 balanced communication and education with respect to the risks, benefits, and responsible use
14 of genetic technologies.

15 With that, thank you.

16 DR. McCABE: Thank you, Dr. Schoonmaker.

17 We're going to hold questions until the roundtable discussion at the end of
18 the presentations this afternoon.

19 Our next presenter is Dr. Ron Bachman, who is Chief of Genetics
20 Department at Kaiser Permanente, Northern California. Dr. Bachman will describe the
21 financing of genetic services in an HMO setting. Having said that, however, I think it's
22 important to recognize that Kaiser is a different model from the typical HMO.

23 Please, Ron.

24 Also, while they're bringing that up, I again will comment that I think it's
25 important to look at the amount of research that comes out of Kaiser in a variety of different
26 areas, because it can be informative in everything from vaccine utilization to genetic services.

27 Dr. Bachman?

28 DR. BACHMAN: Thank you. I'm going to discuss the delivery of genetic
29 health care in a large population, and I'm going to lean to both the clinical and financial aspects
30 of it. The care must be organized, consistent, comprehensive, and cost-efficient, and I might
31 add another C, that of being caring. One must consider the introduction of new technologies
32 and the elimination of unneeded programs. Most programs suffer from an infrastructure that is
33 too small.

34 The problem? Clinical genetics is the health care of a few, where genomic
35 medicine, where we're heading, is the health care of all. Are we prepared? Probably not.

36 I should probably give credit to where I think I cribbed this from, which was
37 an article by someone sitting at the table today, Dr. Collins. Thank you.

38 On one hand, we have the problem of clinical genetics keeping up with new
39 genetic technologies. The genetic evaluation is labor intensive. The current barriers are
40 financial, linguistic, cultural, and many more, and there are a limited number of genetic
41 professionals. On the other hand, we have the problem of the primary care provider. The time
42 needed for a genetic evaluation is quite long. There's limited training in clinical genetics for
43 the primary care provider, such things as genetic testing, risk assessment, non-directive
44 counseling, and psychological implications.

45 I would like to present our program, which we hope will overcome these

1 problems. As Ed mentioned, I work for Kaiser Permanente in Northern California. In Northern
2 California, we have more than 3 million health plan members, more than 4,000 physicians, and
3 more than 34,000 deliveries each year.

4 As I mentioned in my introduction, we think our clinical genetic services
5 are comprehensive, consistent, caring, and cost-efficient.

6 We are fortunate to be well staffed. We have five genetic centers that are
7 strategically located throughout our Northern California region. We have 11 medical
8 geneticists with subspecialty training, four Ph.D. laboratory directors for our molecular and
9 cytogenetics laboratory, 53 genetic counselors, 17 genetic nurses, three metabolic nutritionists,
10 and the support people for the professionals.

11 I'd like to give you an overview of our program. It includes prenatal
12 services in clinical and screening, neonatal services, ethnic screen, multispecialty clinics for
13 common genetic disorders, the adult genetic services which include cancer genetics, clinical
14 genetics, and screening; genetic laboratories and genetic education both for our providers and
15 our members.

16 Probably the centerpiece of our program is our prenatal program. All
17 prenatal patients get a genetic and ethnic questionnaire and a video presentation of our genetic
18 services, which assists them in our informed consent and also the selection of the testing that
19 they think is appropriate for them. There are genetic counselors available for discussing that
20 with them. We also have an expanded alpha fetal protein program, with 80 percent acceptance,
21 an advanced maternal age program. All prenatal patients get an anatomical screen, the so-
22 called ultrasound level 1, which converts to a level 2 ultrasound if any abnormalities or
23 questions are found.

24 We have prenatal cystic fibrosis screening, hemoglobinopathy screening, a
25 fetal pathology program, and genetic counseling when indicated. Our ethnic screening program
26 in the prenatal period includes hemoglobinopathy screening, thalassemia, Tay-Sachs and
27 Canavan, and cystic fibrosis.

28 Our neonatal programs include clinical evaluation. This is for children born
29 with birth defects or that are recognized as being unusual looking. We also have the standard
30 neonatal screening, and we have a program called our Escape Baby Program. This is a
31 computer tracking system for babies that get tested either too early or get out of the nursery
32 before they're tested. We test for the standard four tests that I listed before, and we are in the
33 process of planning a tandem mass spectrometry program.

34 Our screening and tracking programs really include a lot of cases. Last year
35 we tracked over 45,000 cases. These are our prenatal testing, our neonatal testing, and our
36 breast cancer and mammography tracking.

37 In clinical genetics, this is the clinical evaluation by the geneticist, or
38 together with a genetic counselor, or by the genetic counselor alone. Last year we evaluated
39 over 20,000 cases, and this includes case management when indicated, and there's quite a
40 database for outcome studies.

41 The genetic counselors are sort of the glue that holds our program together.
42 They provide genetic services on their own. In fact, they work autonomously in many different
43 genetic type problems. They assist the clinical geneticist in case preparation. They are
44 involved in case management, psychosocial support of the family, and genetic education. It's
45 actually the genetic counselor which is my hope for the future in terms of the integration of the

1 genetic counselor at the primary care level so there isn't a need for the primary care physician to
2 learn all the genetics we are placing on him or her.

3 We also have a fetal pathology program, and we developed this because we
4 were losing important genetic information for diagnosis of conditions and the recurrence risk
5 for those families. Last year we had approximately 2,000 cases.

6 We also have a group of genetic multispecialty clinics staffed by
7 experienced professionals providing care for these particular more common genetic disorders:
8 spina bifida, craniofacial abnormalities, metabolic genetic abnormalities, pediatric lipid
9 disorders, neurogenetics, a skeletal dysplasia clinic, and neurofibromatosis. When a patient is
10 seen in these multispecialty clinics, the recommendations are given both in writing to the
11 patient and their family, and to their provider. Also, our staff in these multispecialty clinics
12 track the patients to make certain that they get appropriate care and that the recommendations
13 that are made are carried out. This is done by either our genetic nurses, our genetic counselors,
14 or our metabolic nutritionists.

15 Multispecialty care is expensive but we think cost-efficient, because the
16 patients are getting appropriate treatment, and that includes appropriate surgery. Last year we
17 had over 2,000 patients in our multispecialty clinics. We also have a cancer genetics program
18 where we do genetics risk counseling, and breast and colon make up the majority of it but we
19 certainly do counseling for other genetic cancers. We provide gene testing when indicated, and
20 as I alluded to before, we have a breast cancer tracking system and a mammography tracking
21 system.

22 The mammography tracking system is for abnormal mammograms. This is
23 not generally in the area that a genetics department works in, but because of our experience
24 with tracking genetic conditions, we were asked to do the breast cancer and mammography
25 tracking, and certainly we thought that the breast cancer tracking system might help us identify
26 some families that had increased genetic risk for breast cancer. Last year in terms of our cancer
27 tracking, we had over 3,000 newly diagnosed breast cancer cases, and over 23,000 abnormal
28 mammograms that we tracked.

29 One needs dependable genetics laboratories, so we've developed our own in
30 cytogenetics, where we've had over 11,000 cases last year; our molecular laboratory, over
31 24,000 cases; and we provide the molecular work for the Southern California Kaiser
32 Permanente. Our metabolic studies go to Southern California Kaiser, where they have a
33 metabolic laboratory. We find it very helpful to manage our own laboratories.

34 I probably, because of time, won't discuss this enough, but we think genetic
35 education and research is very important. We have more experience with the genetic education
36 than research. We provide an online publication for primary care providers which is less than
37 1,200 words. I'm a member of NCHPEG, and when we tried to introduce a CD-ROM that has
38 six hours of terrific information on it, there were a limited number that had the time to review
39 that. So we send out frequent emails, and they're available online, on issues such as
40 hemoglobinopathy screen, who should be referred for cancer genetic counseling, guidelines for
41 management of specific genetic disorders, and guidelines for who should be referred to
42 genetics.

43 We have our own website that was developed, and although I must admit it
44 was developed in terms of education of our members, our providers are using it a great deal so
45 they are prepared for the questions of the members.

1 As I said, we don't have a lot of experience in research. We have the
2 database, we've done a few studies, but not as many as we probably could.

3 Now I would like to talk a little bit about how we introduce new programs.
4 First of all, it's the decision of the genetics group, and those are the geneticists and the genetic
5 counselors. If we think a new technology should be introduced, we prepare a discussion with
6 our new technologies committee, and if they approve it, we then submit it for the Kaiser
7 Permanente budget process. We do this for large programs. We did it when we established a
8 chorionic villus sampling program and when we introduced prenatal screen for cystic fibrosis.
9 We are currently evaluating preimplantation genetic diagnoses and first-trimester screen for
10 chromosomal problems using nuchal translucency.

11 After this process, we submit it to our administration, and if they approve it
12 -- and it usually is approved if it's standard of care and cost-efficient, and we make a good case
13 for it -- we establish a cost basis for the new service, we monitor our productivity and actual
14 costs because the actual costs usually aren't less but may be more than we projected, and each
15 year our programs are reviewed and we have to submit a request for an annual budget. So
16 they're reviewed both at the genetics level and also at the administrative level.

17 This year our budget is going to be over \$24 million, although that includes
18 a \$3 million pass-through fee to the State of California for cooperation with them in the
19 prenatal and neonatal genetic screening program. When I presented this information at the
20 American College of Medical Genetics in 2002, I used our 2001 financial information and
21 figured out that to provide genetic services for our health plan, it cost 52 cents per member per
22 month. For 2004, that's going to go up to 65 cents per member per month, which is a
23 significant increase, but that includes new programs, higher wages of staff, and more staff.

24 Last year we had 207 full-time-equivalent employees in our genetics
25 department. We had 13 physician full-time equivalents. Now, you might remember that I
26 mentioned we had 11 physicians who are geneticists, but we have to employ parts of physicians
27 to help staff our multispecialty clinics, like our pediatric orthopedist or pediatric neurologist,
28 and they charge their time to our genetics program. We saw over 103,000 cases last year.

29 Now, I figured out some cost information that might be of interest to you.
30 Our clinical genetics was \$384 per patient. The multispecialty clinics, as I alluded to, is quite
31 expensive care, over \$1,200 per patient. Our fetal pathology patients were \$270 per case. Our
32 cytogenetics was \$362 per study, and our molecular studies were \$121 per study. The cost
33 seems high, but when you want to provide a quality program and try to provide everything
34 that's standard of practice and include your overhead costs, it certainly is high. Genetics is
35 expensive.

36 WE made an assessment of what we're going to need in the future, and this
37 was done in conjunction with a report from the Health Technologies Center, "Impact of Genetic
38 Testing." We think that in the next two to five years the laboratory needs will be more gene
39 testing, more prenatal genetic screening, more neonatal genetic screening, more carrier testing,
40 more ethnic screening, and more predictive testing. In terms of personnel needs, we're going to
41 need more geneticists, more genetic counselors, more genetic services done by the primary care
42 providers, increased genetic education for primary care providers, increased genetic education
43 for all our residents, and use of the Internet to make genetic services efficient.

44 Now, in terms of the next five to ten years, which I probably should have
45 made five to fifteen years, where we're going seems to be in developing comprehensive genetic

1 services, genetic practice guidelines, preimplantation genetic testing, chip testing for genetic
2 disorders, SNP mapping, pharmacogenomics, and treatment in terms of stem cells, gene
3 therapy, and proteomics.

4 Well, on my last slide I want to tell you what we think the solution is. First
5 of all, a more efficient clinical genetics infrastructure. Even though we have a fairly good
6 system, we are constantly honing it to make it better. As I mentioned before, we need more
7 primary care genetic services, and I am of the belief that we can introduce genetic counselors
8 within the primary care department to work alongside the primary care physicians, to evaluate
9 perhaps genetic history and a pedigree that might have obtained by an Internet use prior to the
10 patient coming into the program, and actually doing counseling for some of the more common
11 disorders such as hemochromatosis, thrombophilia, and cancer at the primary genetic care
12 level.

13 I think we're going to need to incorporate the Internet into genetic services
14 for patient triage, collection of medical history information, pedigree construction and family
15 history, and patient and provider education.

16 We are making some progress on this solution. I think we have established
17 a cost-efficient program and hopefully a model for delivery of clinical genetic services to other
18 large groups of patients.

19 Thank you very much.

20 DR. McCABE: Thank you very much, Dr. Bachman.

21 At this point we're going to take a 10-minute break. We will resume with
22 the next presentation at 3 o'clock. Again, refreshments for members and ex officios are here in
23 the room, and for others in the lobby of the hotel.

24 Thank you very much.

25 (Recess.)

26 DR. McCABE: So next, the next two presentations will discuss the
27 perspectives of the providers. First we're going to hear from Dr. Andrea Ferreira-Gonzalez,
28 Associate Professor and Director of the Molecular Diagnostic Laboratory at Virginia
29 Commonwealth University, who we heard before during the public commentary. Dr. Ferreira-
30 Gonzalez will give us the laboratorian's perspective.

31 DR. FERREIRA-GONZALEZ: Thank you, Dr. McCabe.

32 I would like to thank Dr. McCabe and members of the committee for
33 inviting me here today to share providers' perspective on reimbursement for genetic testing.

34 As has already been alluded to today, codes are the language of
35 reimbursement. They hold the key for laboratorians to get reimbursed or pay for the services
36 that we provide, either by performing the laboratory testing, interpretation or report on that.
37 The current two levels of codes are mostly used by many providers, insurance companies.
38 These procedure codes are, first, Level I. They're called the Current Procedural Terminology,
39 or what we call CPT codes that were developed by the American Medical Association.

40 For laboratories, they usually are five-digit numbers that identify specific
41 analytes, either methodology-specific analytes, assay stains, interpretations, even consultation.
42 There is also another set of codes that could be added to the CPT codes. They're usually called
43 code modifiers, two-digit coded, that are added to the five-digit number that further give a little
44 bit more information about what the depth of the procedure or the interpretation.

45 The second level of coding, the HCPCS codes that were previously

1 mentioned by Dr. Schoonmaker, is the ones that have been developed by the Center for
2 Medicaid and Medicare that allow to deal with testing or interpretation and reports that don't
3 have currently a CPT code approved, or for those newer technologies as they start gathering
4 information to determine what will be the best level for reimbursement.

5 So the use of these CPT codes are the means by which the payers match the
6 service with the appropriate limit of the payment. A majority of the payers, Medicare and
7 Medicaid, all of the private payers, actually recognize the CPT codes to identify the services.
8 In addition to the CPT code, we provide billing or filling out claim forms, we also have to add
9 another specific code that identified the diagnosis or the set of symptoms or signs that are
10 required or triggered the physician to order the different testing, and allows us also to
11 determine what is the diagnosis for that particular patient.

12 For molecular diagnostic testing, there are actually 14 different CPT codes.
13 These 14 CPT codes are used to reimburse for all the genetic testing that is currently used in
14 this country. As you can see here, these 14 codes are procedure specific CPT codes. Here you
15 can see that the numbers are associated with a code from 83890 down to 83912. Each of these
16 different CPT codes has a description associated that allows the third party to identify what part
17 of the procedure, what methodology was used to come up to the diagnosis that we're rendering.

18 As you can see, for example, CPT code 83890 is for molecular isolation or
19 extraction, and 83891 is also isolation and extraction but for highly purified nucleic acid. So it
20 seems that there might be a little bit of flexibility in some of the CPT code they would currently
21 use, but as I'll show you as we go through the different slides, this is not the case.

22 So we have 14 codes to allow us to bill for a large amount of different kinds
23 of services. In addition, all these different services, when they require interpretation and report,
24 we have at different levels. This also is represented by the single CPT code.

25 In here I have provided you the Medicare laboratory fee schedule for 2004.
26 This is the current fee schedule that we use when somebody claims reimbursement for our
27 testing. I'll walk you through this table. In the far corner here, we have the number of the CPT
28 code. Remember the first one I mentioned, 83890 for nucleic acid isolation. As you can see,
29 the first list here is the national limit allowed that Medicare sets for the payment of that
30 particular code. What I don't have here is the lower limit that is allowed, and that is zero. So
31 the national limit goes from \$5.60 down to zero.

32 After the national limit has determined each of the different states, take the
33 code and determine what actually is going to be the level of reimbursement for each of the
34 different states. I have provided you here CPT code and reimbursement with the Medicare fee
35 schedule for different states. In here, this is Georgia, this is Virginia State, and this is
36 Tennessee, North Carolina, and California. What I want to point out here is that even though
37 there is a national limit allowed for all these different tests, there are particular states that
38 reimburse at very low levels for every single CPT code that we use for genetic testing. There
39 are other states that aren't the same.

40 But the other states -- for example, the State of Virginia, where there is a
41 very similar reimbursement level or the national limit allowed, except for one particular CPT
42 code for reverse transcriptase. That is \$17.47.

43 I want you to look at these levels of reimbursement for a couple of specific
44 CPT codes, for the 83890, nucleic acid isolation, and also for the 83891, isolation for highly
45 purified. What you see is a short description of that CPT code, it's implied that it requires

1 further manipulation of the nucleic acid to obtain the more purified that is required for the
2 testing. As you can assume, there is more work, label and reagents that are required to perform
3 this highly purified nucleic acid isolation, but the limit is set to the same level. Please keep that
4 dollar amount in mind as I go through.

5 Before we go through the financial analysis, I think you need to know what
6 kind of services are provided. The Virginia Commonwealth University Medical Center is
7 comprised of three different entities: the Medical College of Virginia Hospitals, MCV
8 Associate of Physicians in private practice, and the VCU Medical School. These are three
9 independent organizations. The Medical College of Virginia Hospital is a 350-bed hospital.
10 It's actually three different hospitals. They are combined under the umbrella of MCV Hospital.

11 We're located downtown Richmond across from the governor's house, so we are downtown in
12 a very large metropolitan city. We serve the central Virginia area of about 850,000 individuals.

13 We are one of the sole tertiary care centers in that central Virginia area.
14 Our laboratory, the molecular diagnostic laboratory, is what's called a comprehensive
15 laboratory. We provide molecular diagnostic testing for infectious disease, oncology,
16 hematology, and inherited disorders. Just to give you an idea of the volume of testing that we
17 handle in the laboratory, in calendar year 2003, meaning January to December last year, we did
18 13,200 tests.

19 Just to give you examples of the levels of reimbursement, I have chosen
20 three example. One example that is highly complex testing and requires highly interpretation is
21 second one, intermediate, and the third one is considered more simple within our standard of
22 high complexity testing. I'm not going to go into detail on Fragile X syndrome because that's
23 not the purpose of this lecture. I'm only going to point out some issues here.

24 Fragile X syndrome is the most common cause of inherited mental
25 retardation, with a prevalence of 1 in 1,200 for males and about 1 in 2,500 for females. The
26 cause of Fragile X syndrome, it's an expansion of the tri-nucleotide repeat sequence comprised
27 of CGG near the 5 prime M of the FMR1 gene. One of the major issues of the diagnosis or
28 trying to identify individuals with Fragile X syndrome is trying to measure the amount of
29 repeats that are located in this area of the 5 prime M of the gene. The number of repeats with
30 allocate the individual within certain categories or areas. If you have 6 to 50 repeats of that
31 particular trinucleotide, you're considered part of the normal population. Individuals with a
32 mutation will have 50 to 200 repeats. These individuals might not necessarily have any
33 phenotype of the disease but are carriers that can pass it along through different offsprings, and
34 actually that mutation can expand to the full mutation that will produce the full phenotype.

35 Full mutations contain about 200 repeats or even higher number of repeats.
36 So there is an area that needs to be very accurate in the amount of quantification of the
37 nucleotide repeats to be able to put individuals within normal populations and at risk or having
38 mutation, and it's about 45 to 55 copies. Due to the fact of the complexity of the sequence and
39 the complexity of the measurement of the repeats, we require the use of two techniques in our
40 laboratory, the polymerase chain reaction and southern blotting analysis.

41 The polymerase reaction allows us to size very good trinucleotide repeats
42 up to 110 repeats. So the normal range plus the mutation. Individuals with larger mutations
43 will not amplify with a PCR reaction. So southern blotting analysis also comes in handy in
44 trying to identify larger permutations and also give us information about the methylation status.

45 So we perform the test, and then we provide the series of CPT codes to the

1 provider to get reimbursed for our test. I have provided here the description and the CPT codes
2 that we currently use for the reimbursement of southern blotting analysis and PCR. As you can
3 see here, sometimes, due to the fact of the way we perform the assays, some of the CPT codes
4 are used more than once. The cost of providing it here is actually the direct cost. It is the cost
5 of the reagent plus personnel. I have not added indirect costs because these will vary from
6 institution to institution. So these will allow you to translate more into different institutions.

7 As you can see here, the total reimbursement for our cost of the testing is
8 \$266.34, and what the Virginia Medicare expects is \$62.30. Remember from that slide where I
9 pointed out to you that the Virginia Medicare expect was only different from the national limit
10 allowed in a single CPT code that we don't use here. So this is very similar to the national limit
11 allowed for this particular testing.

12 For the PCR analysis here, we also have different CPT codes. We do not
13 have a code for nucleic acid isolation because it was already extracted when we performed the
14 southern hybridization analysis. There's a clean-up of the PCR product that is required before
15 capillary electrophoresis, so we needed to add nucleic acid isolation. As you can see here, our
16 cost of performing the test is \$116.06, versus \$17.85. That's what Virginia Medical expect.

17 What I pointed out to you here is that our cost to perform nucleic acid
18 isolation highly purified is \$15.06, and it's about \$55.47 for reimbursement. I also want to
19 point out interpretation and report, our cost is \$40.00 for this other hybridization analysis, and
20 about \$35.00 for that. I want to show you for genetic interpretation, the national limit allowed
21 for all the testing is \$5.60.

22 Now, let's move on to intermediate complexity testing, and I've specifically
23 chosen the immunoglobulin gene rearrangement by PCR because it's also a somatic change. It's
24 not inherited disorder but it's a somatic change, and actually it's intermediate complexity, what
25 we consider complexity in molecular genetic testing.

26 This actually is extremely important in the diagnosis of lymphoma and
27 leukemia because it allows us to identify the proliferation of lymphoid cells, central to the
28 diagnosis of these two entities, because it allows us to differentiate a diagnosis of reactive
29 lymphoidopathy versus lymphoma or lymphoid malignancy.

30 Here again we have a description of the codes that we currently use for
31 seeking reimbursement. As you can see here, we have a nucleic acid extraction of 83891, a
32 highly purified nucleic acid extraction. In the previous slide I showed you that the cost was
33 significantly lower than this one. The reason why this one is so high compared to the highly
34 purified for hybridization analysis is because most of the testing is done on tissue that has been
35 paraffin embedded that needs to be further processed to remove the paraffin and then get
36 washed and get ready for the nucleic acid isolation.

37 What I'm trying to point out to you is that we have two codes for nucleic
38 acid isolation, and I've already described to you at least three different ways to extract the DNA
39 or nucleic acid using different procedures. So there's not much flexibility in the current coding
40 to allow us to account for those differences. In here you will see the particular CPT code we
41 used three times due to the fact that we amplify three areas of the gene or family regions that
42 increase the sensitivity and specificity of our particular assay. Again, interpretation is \$40, and
43 we will get reimbursed \$18.54 because we submit the interpretational report with the 26
44 modifier.

45 But you can see that there's a discrepancy in what it's actually costing us to

1 do the interpretational report and what we were actually reimbursed, and for that matter for the
2 entire procedure.

3 The third example I chose is one of the most simple assays we have in the
4 laboratory and that is widely used and performed in many laboratories. Factor V Leiden is the
5 most common hereditary blood coagulation disorder in the United States. We have a
6 prevalence in the general population of 5 percent for Caucasians and 1.2 percent for the African
7 American population. The reason why it's so important to do genotyping for the Factor V
8 Leiden is that we need to identify individuals that are heterozygous, but mostly homozygous
9 individuals, because we have different consequences of treatment for these particular patients.

10 Being heterozygous for the Factor V Leiden increases your risk of venous
11 thrombosis about five-fold. On the other hand, homozygous it's about 100-fold increase.
12 Individuals homozygous for the Factor V Leiden might be required to place on anticoagulant
13 therapy for the rest of their lives.

14 I have listed here the number of different coagulopathies where Factor V
15 has been associated with. Here again we have the description of a technology and we have
16 nucleic acid isolation, 83890. That's the lowest level of nucleic acid extraction, and you can
17 see the cost is lower too, at \$9.69. This is one of the assays that we perform that has a single
18 amplification technology, currently used in commercially available ASARs have been validated
19 and put together in our laboratory.

20 Again, here we can see that we have to use the same CPT code several
21 times to reflect the procedure that we're currently using. Here, the more simple genetic testing
22 is a lot closer in reimbursement to what it's actually costing us to perform the test.

23 Now, this is how the level of reimbursement. What do we actually get
24 reimbursed? I've showed you Medicare will not be able to provide you specific reimbursement
25 percentage for all the third-party payers of private insurance due to contract agreements and
26 non-disclosure issues that we have with them. But I can tell you that we have submitted a
27 number of claims for all the testing that we did, and we get reimbursement from Medicare
28 about 89 percent of the time, 72 percent for Medicaid, and there's a range for the other third-
29 party payers, from 61 percent up to 85 percent.

30 What it was striking for me to realize that the Medicaid/Medicare
31 reimbursement were getting the national limit allowed. But actually, the third-party payers are
32 paying us almost the identical amount, all of them. So this is something that is across the board
33 for all the different payers.

34 I think also we need to spend some time in the very crucially important code
35 interpretation and report. As we know, interpretation of genetic testing requires the analysis of
36 the testing plus putting information, clinical history, family history, clear pathological
37 correlation, all together to be able to come up with the right result.

38 When we looked at our level of reimbursement for the CPT code 83912,
39 with modifier 26, we see that we get reimbursed, and it's the same level that we get reimbursed
40 for the procedure component of the test that we do, again from 93 percent down to 61 percent.
41 But what we get here, the Medicare/Medicaid gives us the national limit allowed for the 83912
42 with the 26 modifier, but the third-party payers or commercial payers have not recognized the
43 26 modifier and actually reimburse the national limit allowed for the code without the modifier.

44 So what are the factors that affecting access of genetic testing? I have
45 shown you a little bit about the level of reimbursement for the testing that we do, and that has

1 to be crucial to understanding. But also, genetic testing utilization is increasing, and another
2 factor that will dramatically affect access to genetic testing is that the laboratory fee schedule
3 was frozen for five years, from '98 to 2002. After that time, we got a 1.1 increase, and then it
4 was frozen again, and it's going to remain frozen from 2004 to 2008. So we have already lower
5 level of reimbursement, genetic testing will increase, and the fees have been frozen.

6 Now, our costs will continue to increase. Even if reagents don't increase in
7 cost, we're going to have to adjust the cost of living of our personnel, rent, and other expenses.
8 So our ability to cost shift is extremely limited. I'm showing you some examples of this. I
9 think we need to spend some time in this diagram because it gives a very nice example of what
10 is happening in the field of genetic testing and what is to come.

11 The molecular diagnostic laboratory of Virginia Commonwealth University
12 started operation in fiscal year '95. We were doing very little genetic testing. This is a diagram
13 of the different molecular genetic testing that is currently performed in our laboratory, and I
14 have done percent increase through the different years. In 1995, we had barely any genetic
15 testing performed in the laboratory, but you can see that there's a little bit of increase in that
16 particular testing or utilization of that testing. In 1999 and 2000, we've seen an explosion in the
17 utilization of that particular testing. We have gone up to over 100-fold increase in about 10
18 years of service of the laboratory. I haven't put 2004, but I could tell you that it's going to be
19 even higher.

20 So far we have been able to cope with the genetic testing because we've
21 been cost shifting within the laboratory. With the freezing of the Medicare fees, that will put a
22 hamper on our ability to be able to cost shift. What we see specifically is what happened to the
23 molecular CPT codes. When we look at other molecular testing that is currently done in our
24 laboratory and other laboratories, one can see that our ability to cost shift will be diminished.

25 I have given an example here of three different tests. All these tests have to
26 extract RNA, nucleic acid, perform a reverse transcriptase reaction, PCR, and then
27 quantification to quantify the chimeric messenger RNA that is a result of the T922
28 translocation. This is a chronic myelogenous leukemia patient, and this test is crucial for the
29 detection of minimal disease for these particular patients. HIV viral load is crucial for the
30 quantification of HIV in circulation, mostly for patients that are undergoing antiretroviral
31 therapy. More recently, HCV viral load is starting to increase in utilization.

32 As you can see here, we have a little bit higher cost of performing these
33 laboratory-developed assays, and we have a Medicare reimbursement of \$51.65. On the other
34 hand, in the past, the molecular infectious disease CPT code had been reimbursed a little bit
35 better than the other procedure CPT codes. \$99.18 is actually the mean value of cost among 25
36 different laboratories across the country, and you can see the Medicare reimbursement is
37 \$114.36. So we recuperating some of the cost and actually making a little bit. But with the
38 increase in use of hepatitis C viral load and other testing that are not fairly reimbursed
39 currently, these are going to put extra strength in the laboratory and our ability to cost shift will
40 be reduced to almost none.

41 The other factor affecting access to genetic testing that will increase the
42 cost of testing will be royalty payments. Currently we deal with different royalty payments for
43 patented procedures, or even patent genes or sequences. The most common royalty payment
44 for patent procedures is having a percentage fee of the receipts or the reimbursement that we
45 get for the testing that we do, and it can vary depending upon your royalty agreement fee

1 schedule between 9 percent of what you recover to 15 percent of what you recover.

2 On the other hand, royalty payment for patented genes and sequences can
3 have different ways to be performed or different fees, an upfront fee plus a flat fee per test, or a
4 one-time payment plus percentage of the charges. I think it's interesting to see what has
5 actually happened with hemochromatosis in this country. There was a requirement of the
6 patent by SmithKlineBeecham Laboratory, which was acquired by Quest Laboratories, to
7 perform exclusive licensing to perform hereditary hemochromatosis, pretty much testing the 12
8 most common mutations of this particular gene.

9 Quest entered into an agreement with BioRad by which BioRad Laboratory
10 acquired the rights to the patent and developed commercial kits, and it will further sublicense
11 that to laboratories. So you have two options if you want to perform hemochromatosis. You
12 can either buy a reagent from BioRad at a set level or you can license, develop your test by
13 your laboratory, but we have to provide an upfront fee, which is extremely high, plus \$20 per
14 test. You can see what this can do to performing the test for the current cost of performing all
15 the other Factor V Leiden. Also, we can have a one-time payment plus percentage of the
16 charges.

17 The last issue I would like to point out is access to genetic testing, and also
18 our ability to continue to perform testing. It's from work performed by Cho and collaborators
19 that was published last year in the Journal of Molecular Diagnostics, where they did a survey of
20 122 laboratories. The main objective of the study was to try to identify current practices by
21 patent holders and the ability of the laboratories to perform genetic testing.

22 The 122 laboratories were recruited from GeneTests' list and from the
23 directory of the Association for Molecular Pathology. As you can see here, there's a
24 description of the different kinds of laboratories that were enrolled in the survey. It's a little
25 tilted toward university, non-profit private hospital, again because the majority of the genetic
26 tests is currently provided by academic institutions. The companies or commercial laboratories
27 were less represented here.

28 I think what was striking to see from this study is that there was a number of
29 laboratories that received letters from patent holders requesting to stop, cease and desist
30 performing certain testing. There were nine laboratories that received that notice and decided
31 to stop the testing, performing apolipoprotein genetic testing for Alzheimer's disease, and nine
32 laboratories also for breast cancer.

33 I think what was also striking from this study was that 25 percent of the
34 laboratories had to stop testing that they were currently offering. But also, 55 percent of the
35 laboratories expressed that they had not developed a test due to the fact of licensing or patent
36 issues.

37 I hope I've been able to convey to you what is currently happening in the
38 clinical practice of laboratory testing and what some of the major factors are that are going to
39 affect access of this testing to the population.

40 Thank you.

41 DR. McCABE: Thank you, Dr. Ferreira-Gonzalez.

42 Our next speaker will be Dr. Mark Williams for a clinician's perspective.

43 Dr. Williams is a pediatrician and medical geneticist at the Gundersen Lutheran Medical Center
44 in LaCrosse, Wisconsin.

45 Dr. Williams?

1 DR. WILLIAMS: I thank you for the opportunity to be able to speak to the
2 committee. I don't know if speaking or whining is perhaps the operative word here. I'm having
3 flashbacks of dealing with my teenage daughters, I'm afraid.

4 But let me give you a little bit of perspective of a clinician. I'm going to be
5 talking about four different areas, billed services, touch a little bit on multidisciplinary
6 evaluation, access to services, and some issues relating to problems as I see it with the current
7 system.

8 If we look at actual procedures, they really fall into what we truly think of
9 as procedures, which is amniocentesis, CVS, or infusion, and for the clinician what are called
10 evaluation management services or E&M, and I'm going to talk about each of these separately.

11 Procedures themselves are pretty straightforward. The indications are
12 straightforward. You do an amniocentesis for advanced maternal age or for a known
13 chromosomal abnormality, and there are specific CPT codes that cover those particular
14 procedures. Now, that being said, there are of course some insurers that may choose not to
15 cover certain indications, as Dr. Schoonmaker said earlier, and it has to do with exclusion of
16 genetic tests or lack of a benefit.

17 I wanted to spend a little bit of extra time on infusion because there's
18 something new on the horizon here that's going to really impact this not so much for genetics,
19 although that's going to have some impact on us, but particularly hematology, oncology, and
20 some of our colleagues here, and that's the Drug Improvement and Modernization Act of 2004.
21 This defines reimbursement for the infused drugs and biologicals. Essentially, what has been
22 determined is that existing drugs will be reimbursed at 85 percent of the average wholesale
23 price as of April 1st of 2003. New drugs will be reimbursed at 95 percent of their average
24 wholesale price.

25 Well, this has significant implications for treatment of certain genetic
26 disorders where enzyme therapy is now becoming available, specifically Gaucher disease,
27 Hurler-Schie, and Fabry syndrome. Because these are orphan diseases, the enzyme
28 replacement is very expensive depending on the age and size of the individual, anywhere from
29 \$100,000 to \$250,000 a year. It's not an uncommon cost. If the reimbursement is not going to
30 be covering the cost of the medication, that could present access issues for the patients that
31 need these novel therapies.

32 Now, when we talk about evaluation and management, there really are
33 several things that need to be addressed and which I will go through briefly. First is the
34 RV/RVU disparity, and that is that E&M reimbursement is reimbursed at .04 RVUs per minute,
35 whereas procedures are reimbursed at .08 RVUs per minute. So for those of us that don't use
36 scalpels or needles very often, our time is being reimbursed about half of that of our colleagues
37 who do procedures.

38 This is then combined with the issue of how the E&M is actually scored,
39 and I'm going to tell you the system under which we're currently operating and then give you a
40 brief look into a brave new world. The history, it's possible to have a level 4, which is the
41 highest level of a history intensity and not even do a family history. The system as developed
42 really can be done without a family history, and you don't have any ability to bill more
43 intensively for that.

44 Physical examination, again, is based on a number of elements. The
45 elements of the dysmorphology examination, which is very specific, are generally not

1 recognized as elements, although I think it's also fair to say that there are three different
2 element systems out there dating from 1995, 1998, and 2000, all of which are different and all
3 of which can be used to provide documentation to third parties, although depending on the third
4 party, which one they choose to use can vary. And then the third aspect of E&M is the so-
5 called complexity, which is meant to adjust for the seriousness of the condition, risk of
6 mortality, the complexity, but the elements in this part are very poorly defined and are very
7 subject to interpretation.

8 Now, I want to spend a little additional time on family history because that's
9 sort of the geneticist's bread and butter, as you might expect. The standard that we hold
10 ourselves to is a full three-generation pedigree. We frequently are using statistical analysis,
11 including things like Bayes theorem and some of the cancer risk models that are available. We
12 submitted -- "we" meaning the American College of Medical Genetics -- a CPT code, actually
13 two CPT codes for pedigree analysis to define this as something different from family history
14 and use that as a separately billable code to allow us to be able to capture some reimbursement
15 for the complexity of this pedigree analysis that we do.

16 We had an interesting thing happen, which was that just before the vote to
17 approve these was going to be taken by the CPT panel, they said, well, we've got this new E&M
18 system; we need to vote on that and then we'll come back to the pedigree analysis. They
19 approved E&M, which I'll go into in just a bit, and then they said, well, now pedigree analysis
20 falls into the new E&M, so it's off the board. So that was an interesting little end-around there.

21 Well, the new E&M is going to completely get away from the element
22 system that we've grown to know and love. It's going to look at time components, with a
23 maximum time or a level 5 component of approximately 60 minutes. The issue with the new
24 E&M is that the members of the AMA have basically said we don't want the elements, we don't
25 want the documentation, so we're going to develop these clinical scenarios which are really
26 going to define our levels of care. The perspective that I see and that I think Dr. Tunis is
27 probably going to be struggling with in the not-too-distant future is how do you audit that?
28 How do you make sure that people are really doing what they say they're doing? The only way
29 I can really come up with is to audit by time.

30 Well, genetic encounters, as we've already heard, are not infrequently two
31 to three hours face to face. So if we're limited to a 60-minute time, that's going to be
32 problematic, and there's not direct multiplicity of the level 5 versus the level 1. So if I do eight
33 level 1 codes, which I would do if I was wearing my pediatric hat and looking in ears, I can bill
34 much more per hour than I can bill doing one one-hour evaluation.

35 The other thing that is not captured here is the pre- and post-encounter time,
36 and this is something that really has never been built into the CPT system, although there are
37 some modifiers that we do have access to that can take into account pre- and post-encounter
38 time. But as we heard from the previous speaker, it is not unusual at all for third parties to
39 reject these modifier codes, and again the reimbursement does not reflect actual time.

40 Now, there are also codes for coordination of care which we also spend a
41 lot of time doing. Again, these are frequently rejected and do not usually reflect actual time.

42 The issue of physician profiling. This is an audit technique used by third-
43 party payers. It is used to adjust claims to make sure the practitioners are playing by the rules.
44 It's used to adjust charges, and while we're usually told that they can be adjusted up and down,
45 the reality is that they're usually adjusted down. The geneticists have a real problem because

1 we frequently are listed in provider networks -- since we're not able to do a genetics residency,
2 if you will, we do a prior training, usually in pediatrics or internal medicine or Ob/Gyn. We
3 then do our fellowship in medical genetics. We get listed in the network as a pediatrician or an
4 internist, and then we're compared or profiled against pediatricians and internists, and as you
5 might suspect, the code profiles are significantly different.

6 I get information from my pediatric department, and I can tell you that about
7 95 percent of the codes that I submit are level 5 codes, whereas my colleagues, generally 1 to 2
8 percent of what they do are level 5 codes. So when a third-party payer sees that, that can lead
9 to problems saying, well, you're really not doing that, you're just trying to get more money out
10 of us, and can lead to fraud and abuse investigations, or just automatic down-coding.

11 I mentioned the new and improved E&M that will eliminate the elements.
12 These clinical scenarios are going to be developed by specialty societies to define their CPT
13 levels. My current understanding of the process is that there is sort of a beta test of societies
14 that are developing their clinical scenarios to be brought back to the CPT advisory group, and
15 once that analysis is complete, then all societies will be given those as guidelines to go out and
16 do that. Implementation is supposed to be January 1, 2005. Quite honestly, I don't see how
17 that's possible, but that's what they're shooting for. As I already alluded to, the issue of auditing
18 this is going to be, I think, a real nightmare.

19 Multiple providers. We talked about multidisciplinary clinics. Dr.
20 Bachman indicated that it is expensive to provide care that way, although I would argue that if
21 you have an individual that has something like a cleft palate or spina bifida, you're really not
22 going to save any money by having them see the individual practitioners separately. They're
23 still going to have to see everybody. It's a matter of bringing them all under one roof, and so it's
24 an issue of patient convenience. However, there are prohibitions about multiple providers
25 billing on the same ICD code on the same day. The first one in, only one paid is the rule of
26 thumb, so it does encourage some efficiency in terms of getting your bill turned in.

27 Now, in the Down's syndrome clinic where I work at, where we have myself
28 and the developmental pediatrician and speech therapist and others, we have a variety of
29 diagnoses I can use. I use the Down's syndrome code, the developmental pediatrician uses the
30 mental retardation code, so we have different ICD codes that we can use. But if you're in the
31 cleft palate clinic, you're basically stuck with that cleft palate code, and that's going to limit the
32 ability to reimburse because of that ICD restriction. This impairs coordination of care, it
33 inconveniences patients, and it decreases, in my opinion, the quality of care.

34 The last issue relating to multiple providers is the role of genetic counselors
35 and the issue of billable entity status for genetic counselors. The bottom line is that genetic
36 counselors are not recognized as a billable entity, with the exceptions of Washington, Texas,
37 and Ohio, which is pre-HIPAA, which we'll get to in just a bit. Genetic counselors have
38 traditionally not been licensed, although in Utah and California licensure for genetic counseling
39 has been passed. There are restrictions relating to "incident to" billing which are relevant.

40 "Incident to" basically are services provided by a health care professional
41 under the supervision of a physician, which are billed under the supervising physician's UPIN.
42 While I don't want you to get overly concerned about this slide, the first part of it is relevant.
43 This is directly from CMS rules and regulations. "If an employee of the physician provides
44 genetic counseling and that person is not a nurse practitioner, physician's assistant, certified
45 nurse specialist, or certified nurse midwife," -- all four of these having a specific payment

1 recognition in Medicare -- "then genetic counseling can only be billed by the physician as an
2 E&M CPT code 99211."

3 A 99211 is basically office with an established patient, minimal problems,
4 five minutes spent performing. This 99211 was developed for nurses who were providing, for
5 example, immunizations, where they were giving counseling to the parents about the side
6 effects of immunizations. That's what this code is for. But because genetic counselors are not
7 specifically recognized within Medicare with a payment category, they are not able to bill
8 "incident to" at a higher level, as are these folks who can basically have access to all of the
9 E&M CPT codes.

10 Now, there are a few ways that we can sometimes get around it. Hospital-
11 employed counselors can be billed for as part of a facility fee that's not available to non-
12 hospital-based practitioners. Washington State has mandated coverage of genetic counseling
13 and issues billing ID numbers to certified genetic counselors. They are the only state at present
14 that is doing that. I mentioned before Texas and Ohio. Texas and Ohio had also developed
15 private codes, local codes to allow for reimbursement of genetic counselors. However, one of
16 the side effects of HIPAA was that all local codes disappeared. So all of those systems that
17 were developed have been lost to use in those states, and to my knowledge have not been
18 recovered.

19 Even though counselors have licensure status in California and Utah, my
20 understanding is that billing rules are still pending, and so whether they'll actually be granted
21 billable entity status in those states is up in the air.

22 What does this affect? Well, it affects access. Systems don't offer genetic
23 counseling if there's no reimbursement for it. We heard about the labs getting reimbursed at a
24 percentage of cost. Well, the percentage over zero is infinity, so that is a bit of an impediment.
25 Productivity-based reimbursement limits geneticists in the private sector. In other words, if
26 you're in a standard medical group where your reimbursement is specifically related to how
27 much work you do, the amount of work that you're actually able to bill for, given the limitations
28 of the E&M system, really does not make it financially viable in the traditional medical group,
29 although in groups like the group I practice in in Kaiser, where we're on a salary basis, groups
30 can make the decision that this is a value added, and then they'll try and make it up somewhere
31 else.

32 Geneticists are consequently not seen in health plan networks, in the PPO
33 networks that Michele referred to, and there may be referral requirements to see a geneticist or
34 the traditional gatekeeper model. We're seeing a little bit less of that, but I think there's still the
35 perception that genetic services are expensive and are for only the very few, therefore they're
36 really not necessary.

37 Now, I think there are a little bit of things to be hopeful about. The first is
38 that in 2006 we'll all be issued new numbers, thank heavens, national provider ID numbers,
39 NPIs. Our understanding from the interpretation of those rules is that anybody that provides
40 medical services will be eligible to have an NPI. That means that genetic counselors will be
41 eligible to have an NPI. The major problem that genetic counselors have right now is that even
42 if a third-party payer wants to pay a genetic counselor -- for instance, Aetna mandates genetic
43 counseling prior to certain of their cancer predisposition tests -- the genetic counselors can't get
44 into the computer because they're not a billable entity. They can't get a UPIN, they can't bill.

45 If they have an NPI, then certain third-party payers may be able to allow

1 genetic counselors to use that NPI and be allowed billable entity status. There may be some
2 state initiatives that are going to come along. The HCPAC, which is a representative of non-
3 physician allied professionals that sits at the CPT advisory council is developing CPT E&A
4 codes. This is evaluation and assessment codes for genetic counseling to be brought to the CPT
5 Advisory Group and Editorial Panel, and if these are accepted, once a CPT code is in there,
6 basically anybody can use it. So that may help some.

7 Then, as we're going to hear about a little bit later, research on the impact of
8 genetic services on cost and quality of care are out there. There's going to be more of them, and
9 I think we're going to be able to make a better case for why there is a need for our services.

10 Thank you very much.

11 DR. McCABE: Thank you very much, Dr. Williams.

12 Our next two speakers will give us an overview of Medicare's coverage and
13 payment policies and decisionmaking processes for genetic technologies and services.

14 Dr. Sean Tunis, who is Chief Medical Officer, Director of the Office of
15 Clinical Standards and Quality of CMS, will review coverage policies and decisions, and then
16 he will be followed by Dr. Donald Thompson, who is Director of Ambulatory Services at CMS,
17 who will cover payment policies and decisions.

18 Dr. Tunis?

19 DR. TUNIS: Well, thanks very much. I notice that we're running about 15
20 minutes behind, so I'm going to try to focus on just some of the major elements. And I also
21 notice from the last couple of presentations that really all the whining is about payment policy,
22 and that's why I brought a designated whipping boy --

23 (Laughter.)

24 DR. TUNIS: -- Don Thompson. The coverage stuff is really non-
25 controversial, and I'll just whip through it, if that's okay.

26 Just as a broad overview, basically in order to get reimbursed for anything
27 under Medicare, you have to deal with these five bullets, which are regulatory approval, a
28 benefit category determination, coverage, coding, and payment. I'm going to talk about the first
29 three, and then Don will talk about the last two.

30 By the way, I get to present so often with David Feigal of the FDA that he
31 could probably give the rest of my talk, and you're welcome to it if you'd like it. We seem to
32 find ourselves on the same panel about once a week, and I think maybe we'll switch jobs just
33 for fun at some point.

34 Regulatory approval. Basically, you all have heard some of this before, but
35 it's required for Medicare coverage if the technology falls under FDA regulatory purview.
36 Regulatory approval is required for at least one indication, but Medicare has complete
37 flexibility to cover off-label indications for uses of tests, devices, drugs, or anything else, and
38 there's a lot of Medicare payment for things for off-label uses.

39 Obviously, any new guidance adopted by the FDA related to genetic testing,
40 changing the regulatory framework, would affect CMS coverage by virtue of the fact that we
41 would follow that regulatory oversight. On the issue of home-brew tests, since they may not be
42 under the FDA regulatory purview, they do not require FDA approval in order to be reimbursed
43 by Medicare.

44 So really not much more needs to be said about the regulatory issues.

45 The next issue in some ways becomes one of the key issues regarding some

1 of the limitations around Medicare reimbursement for genetic tests, and that's the benefit
2 category issues. Medicare is a defined benefits program, which means we can only pay for
3 things that are specified benefits within the Medicare statute, Title 18 of the Social Security
4 Act. So as examples of defined benefits, there's inpatient services as a benefit category,
5 outpatient services, ambulance services, durable medical equipment. As some of you might
6 have noticed, a new benefit category was added in December of 2003, prescription drugs. So
7 those now become payable by the Medicare program, couldn't be paid for before because there
8 was no statutory basis to do that.

9 Diagnostic services are a benefit category, whereas screening services and
10 preventive services are not. So obviously, the critical issue is what's a diagnostic service and
11 what's a screening service, which turns out to be a rather subtle distinction and an important
12 distinction.

13 So, first of all, this is kind of current policy in Medicare. You have to ask
14 around a lot to actually get this laid out for you. But for purposes of diagnosis, the distinction
15 here is based on whether a person has signs or symptoms of disease or a personal history of
16 illness. So obviously, someone with a history of multiple colonic polyps, they have a history of
17 disease. Genetic screening in that case may be considered coverable.

18 A strong family history would not by itself qualify to make a test a
19 diagnostic test. So genetic testing in high-risk patients with a family history of breast or
20 ovarian cancer would be considered screening, no matter how high the pre-test probability is.
21 It's still considered screening. In some sense, the proof that this is the way the program is set
22 up is that last year we had a discussion about diabetes screening for high-risk patients, and
23 there was a lot of discussion at the Department of Health and Human Services about whether
24 we had the legal authority to do diabetes screening in patients with no signs or symptoms of
25 disease simply based on a profile that would make them very high risk for having diabetes.

26 Essentially, the outcome of that suggests to you what the answer is, which is
27 diabetes screening was just added in the Medicare Modernization Act as a benefit under
28 Medicare. Now, that having been said, our general counsel has long held the view that there is
29 nothing in the statute that actually explicitly prohibits us from designating testing in high-risk
30 individuals with no signs or symptoms of disease and no personal history, nothing that stops us
31 from deciding that that is diagnostic testing. But in order to do that, we would have to go
32 through rulemaking because it's been longstanding agency policy that that's not how we
33 approach it.

34 So if you all were interested in tomorrow deciding what kind of interesting
35 windmills you'd like to approach, one of them would be rulemaking around genetic testing, not
36 that I'm suggesting it.

37 Obviously, any tests that identify a treatment responsive subpopulation,
38 most pharmacogenomic tests, would be diagnostic tests because those are done in patients who
39 have existing signs and symptoms of disease, generally.

40 So that is, kind of in a nutshell, the benefit category issues. Obviously,
41 because of the current situation regarding screening versus diagnostic tests, many of the genetic
42 tests that you all are concerned about are not an issue from the perspective of Medicare
43 reimbursement. We can't reimburse them under the current scenario.

44 Under the situation where tests would fit within the benefit structure and
45 would be potentially coverable, then you move on to the coverage issue, which emerges from

1 Section 1862(a)(1)(A) of the Social Security Act, which says that coverage and payment are
2 limited to items and services found reasonable and necessary for treatment of illness or injury.
3 You've heard a lot of discussion earlier about medical necessity, reasonable and necessary, how
4 that's defined, et cetera.

5 The Medicare program makes coverage decisions at the local and national
6 level. By local level, we mean the Medicare contractors around the country who process claims
7 for Medicare, the local insurance companies. In the absence of a national policy, the policies of
8 those local insurers who process Medicare claims are the coverage policies. Those are called
9 local coverage decisions.

10 We haven't talked about it a lot today, but cost, cost-effectiveness, cost-
11 benefit analysis are not considered formally in making reasonable and necessary determinations
12 within the Medicare program, and the asterisk there is just there to say that's a longer
13 discussion. I wouldn't want to stand up here and suggest that economic issues are not a factor,
14 and I'd be happy to answer questions on that if I'm still around during the panel discussion.

15 So in terms of current situation around coverage policies for genetic testing,
16 we have one national coverage decision, which is on cytogenetic testing, and it specifies it's
17 covered for AML, acute leukemias, congenital abnormalities, and myelodysplasia, I believe.
18 That policy actually dates back to 1979, was updated in 1998, I believe, through the national
19 coverage process.

20 So most other coverage basically, to the extent that there is a test paid for, is
21 under local coverage decisions. A number of the contractors do pay for HER-2 testing. There
22 are some that pay for BRCA testing, although most don't because they consider it to be
23 screening and they are under the same obligatory purview in terms of screening versus
24 diagnostic tests that applies at the national level.

25 So we ran some numbers for 2002. Don's folks did this earlier today. There
26 were 270,000 claims paid at a total cost of about \$13 million. Well, we almost never do a
27 national coverage decision on anything that is less spending than \$50 to \$100 million. That's
28 not a firm or fast cutoff. That just tends to be the way it is, that we're not going to deal with
29 issues that have less impact. So it's likely that there's going to be more national coverage
30 decisions with coming advances in genetic testing, pharmacogenomics and personalized
31 medicine. So the national coverage process may not be particularly relevant to you all now, but
32 it probably will become more relevant as this field advances, particularly if you can get that
33 little thing fixed related to family history of disease and high-risk patients.

34 There is a formal process for developing the coverage decisions by the
35 contractors, the medical directors, carrier advisory committees. There's a development of draft
36 policies, et cetera. So they do have a formal process. There's even a process for reconsidering
37 policies. They basically also apply reasonable and necessary, and I'm going to give you the
38 current definition of reasonable and necessary in a moment. I'd just like to point out that at the
39 local level it's understood that more weight is placed on expert opinion versus empirical
40 evidence. So it tends to be more of a consensus-weighted process as opposed to an evidence-
41 weighted process.

42 So in the context of an earlier stage of development of the evidence around
43 genetic testing, my guess is that there's going to be a lot more tests added at the local level
44 before we ever get any additional tests actually covered at the national level. Local policies are
45 not binding on administrative law judges, and they can be appealed. When there's variations

1 amongst different local coverage decisions in different jurisdictions around the country, that
2 will often be a basis for something being referred up to the national program for a national
3 coverage decision.

4 This is the diagram of the national coverage process. Again, we could
5 spend a long time on this, but basically this is just to emphasize that there is a formal
6 articulated process. This is in the Federal Register written out just this way. It involves the
7 possibility of technology assessments being done, usually through the Agency for Healthcare
8 Research and Quality. We have a Medicare Coverage Advisory Committee, and the time frame
9 for this is now six to nine months. These are new time frames that were imposed as a result of
10 the Medicare Modernization Act. We had target time frames in the past which we applied
11 rather loosely. So anyway, now there's a nine-month time frame during which a decision had to
12 be made.

13 So here's the definition from Medicare's point of view of what's reasonable
14 and necessary. There needs to be adequate evidence to conclude that the item or service
15 improves net health outcomes, and in terms of health outcomes we emphasize outcomes that are
16 actually experienced by patients. So functional status, quality of life, psychological outcomes,
17 as well as morbidity and mortality, all of those are meaningful outcomes when it comes to
18 evaluating the impact of a diagnostic test.

19 We usually look for evidence that's generalizable to the Medicare
20 population because there are sometimes significant differences between the performance of the
21 technology, including genetic technologies I imagine between younger and older populations,
22 and we also look for things that are as good or better than currently covered alternatives. We
23 use a standard evidence-based framework, the same sources of evidence as I think Michele
24 Schoonmaker put up, published literature, systematic reviews, expert guidelines, et cetera.

25 The key factor here is that we weight the evidence based on the source and
26 the methodology. The whole evidence-based medicine approach basically says you put more
27 emphasis and place more weight on evidence from sources that have less potential for bias.

28 So let me just talk, then, about determining the adequacy of evidence related
29 to diagnostic technologies. We use a framework which is not unfamiliar to anyone here, but it
30 was articulated by our Medicare Coverage Advisory Committee, that basically there are sort of
31 two critical pieces. There is test performance, the sensitivity and specificity or accuracy of the
32 test, and then there is the clinical utility, impact on patient management and outcomes. In some
33 cases, of course, the clinical utility will depend on there being a beneficial intervention
34 available.

35 So then we come down to an important question, which is under what
36 circumstances does information itself provide benefit? Well, we certainly don't assume that
37 information itself and increased certainty is inherently beneficial. At least, I'm not aware that
38 that's an assumption that we've been using.

39 So the value and the impact of the information is likely to vary by the test
40 and by the specific clinical circumstances, and what I would say is that ideally, if one is going
41 to argue for the benefit of information, whether it's a Huntington's disease test that presumably
42 affects people's lifestyle choices or other factors, we would look for some evidence to
43 empirically demonstrate that the information in fact has that impact. Simple qualitative
44 arguments that that might be the benefit of a test are not going to be sufficient, at least at the
45 national level, to merit coverage.

1 So that covering the regulatory issues, the benefit category issues, and a
2 quick intro on reasonable and necessary, and we'll let Don take all the hard stuff.

3 DR. McCABE: Thank you.

4 Mr. Thompson?

5 MR. THOMPSON: I was sitting in the back listening and taking an interest
6 in Dr. Ferreira-Gonzalez' presentation, and it was so good, about halfway through I started
7 thinking what idiot is in charge of the Medicare physician clinical lab fee schedule? And I
8 realized that was me.

9 (Laughter.)

10 MR. THOMPSON: So not considering myself an idiot, I'm going to talk a
11 little bit about how we do new determinations for payments, and then a little bit about how we
12 got to where we are, because when I first took this position and I looked at the Medicare
13 clinical lab fee schedule, I came to many of the same conclusions that some of the earlier
14 presenters have come to. You have to dig a little to kind of understand the tortured history and
15 kind of understand a little bit about how we end up in a position where you have some of those
16 slides that were presented earlier that have such payment anomalies surrounding them.

17 But first, let me talk a little bit about how we handle new lab tests. We have
18 two methodologies primarily for handling payments for new clinical lab tests under the
19 Medicare clinical lab fee schedule, gap filling and cross-walks.

20 By the way, everything I'm saying here applies to all new technology under
21 the Medicare clinical lab fee schedule. We don't have anything specific to genetic
22 technologies.

23 So gap filling and cross-walking. Gap filling is a process where we
24 essentially go to all of our carrier medical directors, all the medical staff in our contractors, and
25 we say we have this new test that CPT has created, because the beginning of this process is a
26 new CPT code. That's how this comes about. So we get a new CPT code that gets created, we
27 go to our contractors, and we say we'd like you to kind of go through, examine this test, and
28 look at a variety of factors that I'll touch on in a later slide, and determine a payment amount for
29 your area. That's the gap-filling process.

30 The cross-walk is done in Central Office. So in cross-walking, what we say
31 is we have a new lab test and a new CPT code, and we think that this is similar in certain
32 respects to an existing test. We just cross-walk the payment amount to an existing test.

33 So those are the two ways. Gap filling, it's a decentralized process where
34 we send it out to our carrier medical directors. Cross-walking we do in Central Office where
35 we look at what existing tests under the clinical lab fee schedule might be appropriate and we
36 cross-walk the payment to that.

37 We don't do this in isolation. We have public meetings. We have public
38 input on this. The one for 2005 will be on Monday, July 26th, for those of you that are
39 interested. Any interested parties can give testimony. The test kit manufacturers can come in.
40 ACLA can come in. It usually comes in. AdvaMed gives presentations. Everyone gives us
41 recommendations about how they think we should either gap fill or cross-walk and, in the
42 situation of cross-walk, what code they think we should cross-walk to. We then take these
43 tentative determinations and we post them on the web for additional public comment, and we
44 make the final determinations usually around the early part of November, and that would be for
45 the 2005 tests.

1 Gap filling. Let me say at the outset I'm not a huge fan of gap filling. It is a
2 great process in concept, not fantastic in execution.

3 What we say on gap filling is a carrier should examine a variety of factors,
4 and we do not weight these in any way. These are just kind of guidelines for them. Charges for
5 the test, routine discount to charges -- so look what the lab is charging for it -- and look at the
6 resources required to perform the test. Look what other payers are doing in your area, although
7 as an earlier presenter mentioned, that's somewhat circular because to the extent that they're all
8 keying off the clinical lab fee schedule, it's hard to look at what they're doing because they're
9 waiting to see what we're going to do. Then charges, payment amounts, resources required for
10 other tests that may be comparable or otherwise relevant.

11 Then in addition to kind of those core items that they look at, it's also
12 clinical studies and information provided by clinicians practicing in the area. They obviously
13 have a network of physicians that they can tap into. Manufacturers and other interested parties
14 are allowed to submit comments to each of the carrier medical directors when they're making
15 their gap-fill determination.

16 So kind of against this backdrop, which is how we've been doing it for some
17 time, which is this gap filling versus cross-walking methodology, you have the Medicare
18 Prescription Drug, Improvement, and Modernization Act of 2003. There were two primary
19 provisions in there that affected the clinical lab fee schedule and more of a secondary one that
20 might have some long-term impacts, but let me talk about these two first.

21 As an earlier presenter mentioned, no updates to existing lab fees until
22 2009. That is a blanket freeze. There is no administrative discretion there. So there's not a
23 whole heck of a lot we can do at CMS in terms of adjusting lab fees for existing tests until
24 2009. So some of those slides that you saw earlier that had some of those payment anomalies
25 in will continue to exist for some time, and there are many stories in the clinical lab fee
26 schedule like that. That is isolated to genetic testing by any means.

27 Then the other section, though -- there is a small ray of hope here -- in
28 942(b), Methods for Determining Payment Basis for New Lab Tests, it goes into a little bit
29 about the need for us to publish regulations in this area. We have not had regulations in the
30 past. Most of the payment methodologies under the clinical lab fee schedule have just been a
31 past practice. They've kind of arisen over time and they have kind of self-perpetuated, but we
32 do not have formal regulations, and our general counsel would indicate our past practice has the
33 force of regulations. If we have an established process, we can't just say, you know, we think
34 next year we're going to change the whole thing around. So the fact that we have past process
35 means that if we want to change that, we have to go through a regulation and now what Section
36 942(b) does is say, okay, we're required to go through a regulatory process.

37 The odd thing, though is when you look at what Section 942(b) says, it
38 describes to a certain extent what our current process is. So it didn't give a lot of guidance in
39 terms of what Congress might have intended for us to under 942(b), other than one could
40 envision the kind of process that we were currently using, which is get public input, the cross-
41 walking, the gap filling.

42 So we're struggling with -- my last point there, differences from current
43 process -- we're struggling with this and what we intend on doing is going through a formal
44 notice and comment. So we will this year have a formal regulation, a proposed rule that we're
45 going to put out, and we'll kind of go through what we think our interpretation of 942(b) is and

1 seek public comment on that. I'm very interested myself in seeing what kind of comments we
2 get, and then we'll go through a final rule process after that.

3 So the issues for reform, we touched on the relative payment rates are
4 essentially frozen, and this is for two reasons. Not only the explicit freeze in the MMA, but in
5 addition, going into some of the history of the clinical lab fee schedule, it is the oldest fee
6 schedule in the Medicare program. It dates back to the early 1980s. It is very archaic at this
7 point.

8 However, there's no mechanism for revising it. There was some optimism
9 that perhaps in the recent law we would get authority, that CMS would get authority, to revise
10 the clinical lab fee schedule in its entirety. I mean, not just for genetic testing, but in the whole
11 thing, look at it comprehensively and say what do we have here? We have a system where the
12 clinical lab fee schedule was established in the early 1980s and life is a lot different now, but
13 yet we still have these same relatives locked in place from the early '80s and it's difficult to get
14 at them.

15 In addition, maybe give us a different methodology for adding new tests.
16 The 942(b) was helpful in that respect, but it would have been nice maybe if Congress had
17 more explicitly provided some guidance, but we will struggle through that from a regulatory
18 standpoint.

19 To a certain extent, going back to some of the earlier slides, this is where
20 the problem is. You have this 1980s fee schedule that we've attempted to kind of modify over
21 time, even though the basic construct stays the same and we don't have any statutory authority
22 to revise it. We've tried to modify it over time to make it work for new lab tests.

23 But as you can imagine, one of the immediate issues that comes about, think
24 about cross-walking. Okay, you have a new test. You say this is kind of like this existing test,
25 but the payment rate for the existing test was locked in in the early 1990s. So you're in a
26 situation where the logical choice for the code -- you say okay, this definitely walks right to this
27 code, this is similar, and this is how you should pay it, but the payment amount for that code
28 doesn't necessarily make sense. So you're kind of in a bind.

29 So at that point, what do you do? You say, okay, we'll throw it out to gap
30 filling and see what happens there, but because of the decentralized nature of that, sometimes
31 the proponents of the CPT code can be nervous sometimes about going out to that gap-fill
32 process.

33 It's a little bit of a Catch-22. You're cross-walking to what may be a
34 mispriced code or you're sending it out to gap filling, which is a decentralized process where
35 you have to deal with all our individual carrier medical directors.

36 So where does that kind of leave us? Not in a great spot. One of the things
37 that we'd like to look at is there was an Institute of Medicine report that talked about revising
38 the clinical lab fee schedule and they had a lot of excellent thoughts and suggestions about how
39 we might go about doing that. Now, we have no statutory authority to do it, but at least we
40 might make some more progress thinking about those thoughts.

41 Along those lines, one of the recommendations in that report was to look at
42 kind of a competitive bidding process. For those of you familiar with other sections of the
43 MMA, there is competitive bidding now for durable medical equipment, there is competitive
44 bidding for Part B injectable drugs that will be coming up in 2006, and there is a demonstration
45 project to do competitive bidding for labs. So that's one concept one might use in thinking

1 about revising the lab fee schedule in total, is that kind of competitive bidding approach.

2 Another one might be negotiated rulemaking. That's worked successfully,
3 for example, on the ambulance fee schedule. That's another road we might go down.

4 There was some optimism about looking at charges for this. For those of
5 you familiar with the kind of Tennant fiasco and some of the problems we had on outlier
6 payments, that's thrown a little cold water on the concept of using charges, but it still might be
7 something worth looking at.

8 So those are kind of the options out there, but again, no statutory authority
9 to implement them. The only thing we can do, with respect to some of the older technology
10 tests, we do have an authority in the statute, inherent reasonableness authority. What we're
11 allowed to look at there is things that are inherently unreasonable, payments that are inherently
12 unreasonable, and there's a process in the statute.

13 There was a moratorium on using that authority. That moratorium has been
14 recently lifted, and one of the things we're examining very early in that process, once we issue
15 instructions to our contractors, is in fact the HIV/HCV viral load that was mentioned earlier,
16 where you have the payment rate for the HCV load is roughly half of the HIV for what is
17 almost the identical procedure.

18 So that's an early candidate for us, is inherent reasonableness. It may be
19 difficult to revise an entire fee schedule brick by brick, but the inherent reasonableness
20 authority is one avenue we could go down and we're interested in using that. Hopefully, this
21 year we'll have those instructions out and be able to at least start with some of the more
22 egregious examples.

23 I can take questions during the roundtable.

24 DR. McCABE: Thank you, Mr. Thompson.

25 Now, Dr. David Veenstra will discuss cost-effectiveness analyses of genetic
26 technologies and informed coverage and reimbursement decisions and the data needed for
27 conducting such analyses. Dr. Veenstra recently joined the University of California-San
28 Francisco School of Pharmacy and is an assistant professor in the Department of Clinical
29 Pharmacy.

30 Dr. Veenstra?

31 DR. VEENSTRA: Thanks to the committee for inviting me to come and
32 present today.

33 The other speakers I think really gave a great background for talking about
34 cost-effectiveness. I just want to give a little bit of an overview of cost-effectiveness, kind of
35 the 20,000-foot level, talk a little bit about economic evaluations of genetic technologies, and
36 then go over some examples to kind of give you an idea of what types of information you can
37 get out of these types of analyses.

38 I guess the kind of message I want to get across is that cost-effectiveness
39 analysis is not about just looking at cost only. It's really about putting a value on all of the
40 things that we look at in health care, including quality of life and life expectancy, and really I
41 think the advantage of formal cost-effectiveness analyses is that they provide a framework for
42 evaluating the complex and conflicting factors that are involved in making coverage and
43 reimbursements decisions in health care, and I think we all realize that with genetic
44 technologies that's even more of an issue.

45 Another advantage that you can have here, and we'll see this as we go

1 through the examples, we can look at multiple different strategies and many times, when we use
2 modeling techniques, we can get information to decisionmakers when they're trying to make
3 that decision, as opposed to years later.

4 I'm not going to go into all these different details here, but I just want to
5 highlight that within this kind of area of economic evaluation in health care, there are a variety
6 of different methods that folks use. A lot of times these are all referred to as cost-effectiveness
7 analysis in general.

8 I think really kind of the gold standard in the field is what's called cost-
9 utility analysis, and the reason that basically people like it is that you look at outcomes in
10 quality-adjusted life years. So you look at life expectancy as well as the impact on quality of
11 life in patients, and of course, costs are measured in typical dollar terms.

12 So one of the questions that was put to me was what information does cost-
13 effectiveness analysis provide to health plans? Well, unfortunately, it's just one little piece of
14 the puzzle, and I think as we've heard from the discussions today, there are a tremendous
15 number of issues that go into decisionmaking. So I don't want anyone to feel like I'm
16 advocating using cost-effectiveness analysis to make decisions about what to cover. It's just
17 one piece of the puzzle, but I also think that it can highlight some strengths and weaknesses in
18 these other areas.

19 Another tough question is is cost-effectiveness information used in
20 reimbursement decisions in the U.S.? Well, I think we heard one perspective from Sean, and I
21 can bring a little bit of another perspective on this issue, more from the managed care arena.
22 The short answer to that question is sometimes, and it's changing.

23 I think a nice example that's out there of the use of economic information in
24 decisionmaking are the guidelines that have been put out by the Academy of Managed Care
25 Pharmacy. These were put out and approved by the board of directors in October of 2000, and
26 basically the objective of these guidelines for making decisions about pharmaceuticals were to
27 improve access, improve the transparency of the information of the decisions, and try to
28 achieve a consistency in making decisions, cover and reimbursement decisions about drugs to
29 put on formularies.

30 Basically, what this format does is the managed care companies go to the
31 manufacturers or the pharmaceutical and biotech industry, and they say give us all the
32 information you have on your drug, including unpublished studies, and also provide us with
33 evidence of cost-effectiveness.

34 So it kind of provides a framework for looking at this and it enables, when
35 these decisions are made and when folks are making decisions in managed care, instead of just
36 looking at one or two or three studies or the ones that have been published, you're able to have
37 access to all of the information that's out there. There's obviously a lot involved with checks
38 and balances and making sure this information is accurate and exhaustive, et cetera, but
39 basically, it ends up giving the decisionmaker more information with which to make that
40 decision.

41 So cost-effectiveness is a piece of that. The rest of it is really based around
42 evidence-based decisionmaking.

43 This I think is about six months old, but the utilization of this format has
44 become fairly popular. A lot of managed care organizations across the U.S. are adopting it,
45 including a few of the states, and it's really become kind of -- hopefully, it's not a trend. It's the

1 current kind of gold standard I think in managed care. There's a lot of other folks out there that
2 have been doing this type of thing for a long time and doing it very well, such as Kaiser, et
3 cetera, but there are a lot of smaller plans out there that really haven't been adopting a formal
4 evidence-based evaluation as well as cost-effectiveness.

5 Just in our experience with running training programs, et cetera, when do
6 you actually use cost-effectiveness information? Well, it's generally when you have, for
7 example, more than one drug in a class of drugs and you're basically making your decision
8 based on price, and so you may be wanting to look at things such as the cost of side effects and
9 monitoring things like that.

10 I think the area where it draws the most attention is when you have an
11 expensive and novel technology. So maybe, for example, when Entercept came out for the
12 treatment of rheumatoid arthritis. This is probably the situation where some genetic
13 technologies are going to find themselves and this is when payers start to get interested in
14 issues of cost-effectiveness.

15 This issue of do payers care yet, I think it's not quite on their radar screen
16 yet. I think when you talk to them about genetic testing and pharmacogenomics, a lot of folks
17 start to think, "Oh, biotechnology, biologics. Big budget impact. Now I'm concerned." So if
18 it's something that they feel is high tech and expensive, they'll start to get interested.

19 I think in some of the data that Sean showed, it's not really having a big
20 enough impact quite yet where at least folks in, for example, managed care have it at the top of
21 their list. I think once we start to get more common genetic testing or tests that really influence
22 the utilization of expensive drugs, you're going to start to see a lot more interest in cost-
23 effectiveness of genetic technologies.

24 That's basically what I've covered here. Also, obviously, with regulatory
25 intervention there would be greater interest also.

26 So what are the determinants of cost-effectiveness of genetic technologies
27 and what makes something cost-effective? I just want to flip through a couple of issues here,
28 just some work I've done with colleagues thinking about what could be important.

29 Really, for pharmacogenomics, there's a lot of information out there about
30 drugs where their metabolism is influenced by genetic makeup in terms of drug-metabolizing
31 enzymes, but you really have to ask yourself how serious are the side effects? Are they going
32 to have significant patient impacts and economic impacts? If you're using pharmacogenomics
33 to select a drug, is that an expensive drug? Is that something that's used over the lifetime of a
34 patient? How much money are you going to save?

35 The same thing with disease genetics, looking at disease risk. You really
36 have to consider what are the ultimate outcomes you're trying to prevent.

37 Another issue that I think that I've especially felt that in pharmacogenomics
38 is often ignored is the concept of what's the next best thing? What's the comparator? What's
39 the alternative?

40 So are you going to use genetics to decide which drug to use for a patient to
41 treat their hypertension or do you just have them come back into the office every few months
42 and over a six-month time period or a year time period, you get it figured out which drug works
43 for them and which dose? That may not be too expensive, so paying a lot of money for an
44 expensive test may or may not be worth it.

45 Of course, I think this audience here is familiar with the issue of association

1 studies on the correlations between the genotype and the phenotype. A lot of the folks out there
2 I've seen will focus in on test sensitivity and specificity as opposed to considering actual
3 clinical outcomes in the patient and actual phenotype. Obviously, genes that have a higher
4 penetrance will be more cost-effectiveness in terms of testing.

5 There are issues around the cost of the test. I think the presentations here
6 today have covered a lot of this. There are things that we need to consider, such as the induced
7 cost and additional clinic visits.

8 I think there are some benefits and potential benefits in genetic testing that
9 does differentiate it from other types of diagnostics in terms of the ability to use that
10 information throughout the lifetime of the patient. For example, this Roche amplichip, which
11 looks at a series of drug-metabolizing enzymes. Basically, you can look at your entire drug-
12 metabolizing profile on one chip. For instance, if you got that test when you were a young
13 child, you'd have that information available to you for the rest of your life, and the cost is really
14 kind of alleviated that way.

15 The last point is basically that genetic testing is essentially a screen or
16 preventive medicine, and for those of you that work in that area, you know that cost-
17 effectiveness is highly driven by the prevalence of the underlying disease, and in this case that
18 would really be the prevalence of the genetic variant. If the prevalence of a genotype is only
19 0.5 percent, we have to go and test 200 people. So obviously, this is going to have a big
20 impact.

21 We did a hypothetical cost-effectiveness evaluation of testing the TPMT
22 gene, which Michele mentioned earlier, for childhood leukemia treatment with 6-
23 mercaptopurine, and we basically used decision analysis and built a decision model in terms of
24 whether you would test or not and whether the patient was deficient or not and whether they
25 had a serious adverse drug reaction and there is potentially mortality associated with that.

26 Now, this was just an example, so we kind of tripped this thing out, but
27 basically we put in some parameters for the cost of the test, the mortality due to the adverse
28 drug reaction, and then the prevalence of the deficient genotype. We found that these three
29 parameters represent three of the dimensions in cost-effectiveness. The cost of the test, the
30 economics; the genetics, the prevalence of the genotype; and kind of a clinical outcome.

31 What I'd like to show you on this graph here is basically what we've done
32 here is plot -- this is the mortality associated with the adverse drug reaction, that's the cost of
33 the test on this axis, and on this axis here we have what's the incremental cost-effectiveness
34 ratio. Basically, just to make this simple, things that cost less than \$50,000 per quality-adjusted
35 life year in the field is somewhat considered reasonable and cost-effective. When you're
36 between 50 and 100, not quite so. It's more of a question mark, and then above 100, it's
37 generally not considered cost-effective.

38 So in this example, when we're using the default prevalence of the genotype
39 of 0.3 percent, you can see that depending on these other parameters, you may or may not be
40 cost-effective. It may not be a reasonable way to spend your money compared to other
41 interventions that you could be allocating your budget to.

42 But when we change this from just 0.3 percent to 1 percent, you can see the
43 significant impact it has on the cost-effectiveness. So even small changes in the prevalence of
44 the genotype can have a big impact, even in terms of budget impact in terms of the number of
45 people that you identify.

1 Now, I just wanted to give you guys a few examples from the literature.
2 This is a study published recently, November 2003, looking at newborn screening for MCADD,
3 and this was using a tandem mass spec technique. Just briefly, basically what they did was they
4 created a cost-effectiveness model using decision analytic-type techniques, like I just showed
5 you there, and they did a cost-utility study. So they were looking at cost per quality-adjusted
6 life year.

7 They assumed the cost of the test was only an additional \$4. This was
8 assuming that there was already some type of tandem mass spec testing going on, and they
9 looked at what would happen with screening versus no screening in looking at the 2001 birth
10 cohort.

11 Well, they found that you'd end up with longer and better lives. So you'd
12 have a gain of 990 quality-adjusted life years. So let's just say 1,000 life years is what you'd
13 save.

14 You would have, however, a higher overall cost of \$5.5 million, but if we
15 convert that into the cost-effectiveness language, we end up with \$5,600 per QALY, which is a
16 lot less than the \$50,000 per QALY. So this might tell a payer that this is a reasonable
17 technology to reimburse.

18 If we look at colon cancer, there's been some work in this area. Scott
19 Ramsey has looked at testing for HNPCC, and again, I think Sean outlined this nicely.
20 Basically, what they found is that the incremental cost per QALY is about \$40,000 per QALY
21 if you look only at the patients, but when you include family members, the siblings and children
22 of those patients, all of a sudden you get tremendous more gain for the amount of money you've
23 spent. I think that might have interesting implications in terms of who's covered and who's
24 reimbursed for genetic testing and obviously can run into some difficult reimbursement issues.

25 I think there is also some interesting information that's come out of some of
26 these studies where some folks have advocated universal screening as opposed to folks that fit
27 within the Bethesda guidelines, et cetera, and have found that the cost, the annual cost, in the
28 United States could be pretty dramatic if we were to end up full testing on all patients.

29 Breast cancer. There's been not as many studies as you might think of the
30 cost-effectiveness of actual screening, but Grann and colleagues have a study looking at
31 Ashkenazi Jewish women, and again, not to go through all the details of the disease, basically
32 what they found was that for a cohort of 10,000 women, you could avert about 80 deaths by
33 having this screening program. They found in their calculations that you were doing this at a
34 rate of about \$30,000 per life year saved, so something that seems very reasonable.

35 However, they conducted that analysis with a default value for the test of
36 about I think \$400, and when you plug in something perhaps a little closer to what might be
37 reimbursed, now you're up in the \$85,000, pushing \$100,000, per QALY. So a payer might
38 utilize an analysis like this to give some pushback on the cost of a test or try to negotiate on the
39 cost of a test based on its value.

40 Now, does this happen every day with the Academy of Managed Care
41 Pharmacy guidelines? No. It's a much more subtle effect, I think, but it provides a framework
42 for these types of negotiations.

43 Then lastly, for pharmacogenomics, this is some work that Katherine
44 Phillips, a colleague of mine from UCSF, has led. It's actually working with the FDA, doing
45 some work with them. She did a systematic review looking at cost-effectiveness studies of

1 pharmacogenomics.

2 Basically, just to sum up, she found 10 studies out of a total of 253 citations
3 that were identified. Four were in thromboembolic disease, a couple were in chronic hepatitis
4 C, two were looking at the enzyme thiopurine methyltransferase, and a couple in other areas.
5 Eight found that genotyping was relatively cost-effective, while two studies found it to be less
6 cost-effective than other options. So that's kind of the landscape of what's out there for
7 pharmacogenomics.

8 I'll skip this slide.

9 So what are some of the unique challenges of looking at cost-effectiveness
10 of genetic technologies? I think a lot of it is fairly obvious. Basically, there's a lot of
11 information, it's complex, and it's interacting, and so there are pretty significant data needs.

12 I think some of the things that need to be worked out are what are really the
13 induced costs surrounding testing. A lot of these issues of the cost of adverse drug reactions,
14 for example, I don't think have been costed out very well, and the whole issue I think of patient
15 preferences and quality of life needs to be looked at a little bit more closely.

16 You know, I feel that using a decision modeling framework, you can kind of
17 bring all these complex factors together and with additional data in these areas in the economic
18 outcomes, the patient outcomes, as well as obviously, as the committee discussed earlier, the
19 clinical outcomes and association studies, with this type of information I think it is possible to
20 provide useful cost-effectiveness information to decisionmakers.

21 So along the lines of what I think everyone's been talking about and
22 thinking about are providing some types of guidelines and policies for the reimbursement, using
23 an evidence-based approach, and incorporating some aspects of cost-effectiveness into the
24 process.

25 I think some issues that I've seen will be who will be responsible for these
26 decisions. Is it going to be the P&T committees or medical services? I think for
27 pharmacogenomics, it may very well be that it falls under a P&T pharmacy services area.

28 I think there's particular interest with regard to pharmacogenomics in terms
29 of it's a real pain for them to try and control drug use by requiring prior authorization, but if a
30 drug comes with a genetic test that's required, it kind of makes them easier to control drug
31 utilization and expenditures potentially.

32 So in summary, I think cost-effectiveness evaluations in health care is
33 challenging to begin with. I think in genetic technologies, it's probably one step further.

34 I think because, however, the decisions are so complex and there are so
35 many factors involved, that decision analysis and cost-effectiveness analysis at least provide
36 you with a framework and it can highlight where your data uncertainties are and where
37 additional resources need to be invested.

38 I think that as more tests come to market, that the need for these types of
39 studies are probably going to be increasing as payers are faced with more decisions in this area.

40 So that's it. Thanks.

41 DR. McCABE: Thank you very much, Dr. Veenstra, and thank you to all of
42 the presenters for these very informative and helpful presentations. Now we can explore these
43 in a roundtable.

44 Just while you're sitting down, Dr. Veenstra, you talked about the
45 complexity of these issues, and one point that wasn't raised, but one of our fellows presented

1 some work at the Western Society for Pediatric Research and has a manuscript in preparation.
2 What he found -- this was in a study of newborn screening for severe combined
3 immunodeficiency, SCID -- was that it was most sensitive, looking at newborn screening, to the
4 sensitivity and specificity of the test to the test parameters because that determined the false
5 positive rate and those sorts of things.

6 So there are even additional issues, and that analysis was much more
7 sensitive to those issues than to the frequency because of the cost implications in a screening
8 test. So I think these are just incredibly complex issues as we move forward and it will depend
9 on the setting in which the testing is performed as well.

10 Now it's time for the committee to ask questions and make comments about
11 the presentation. Yes, Joan?

12 DR. REEDE: One of the things that was very striking to me was bringing
13 home a clear recognition that as we talked about the need for more evidence and the challenges
14 that we're facing or going to be facing for those who are insured is the widening gap for those
15 who are not insured, and the fact that these issues just sort of see us moving toward a
16 potentially two-tiered system, that as we move towards advancing technologies and use of these
17 technologies and trying to figure out how to pay for them, there is a huge portion of our
18 population where these will not be accessible.

19 I'm wondering, from any of the presenters, if you could give any comment
20 to what might be done in terms of addressing this part of the population that has been left out of
21 this discussion.

22 DR. McCABE: Members of the panel? Marc?

23 DR. WILLIAMS: I'll take a crack at it. I mean, I don't see anything
24 different here than what we're facing in medicine in general. I mean, the issue is we have a
25 two-tiered system. We have rationing. We may not want to use those terms, but that's the
26 reality. We have a limited budget and limited resources, and so in some sense it does seem a
27 bit silly to be arguing about who gets what when we know that there's a large population of
28 people that probably don't have access to much of anything. But I don't know that there's
29 anything specific to the genetic piece that separates that out from that issue. Pick any sort of
30 access to service.

31 I can tell you that in my own clinical practice, that because of the nature of
32 genetic disorders, that a lot of these children and adults do qualify for coverage under some of
33 the special needs pots of money that are somewhat separate from other Medicaid pots, KBAC
34 and other of those sorts of funds.

35 So at least in Wisconsin, we haven't had a tremendous issue with children
36 and adults affected with certain genetic disorders in terms of getting them qualified, and thus
37 eligible for services. So I think that in some sense having a genetic disorder may actually make
38 available some reimbursement systems that aren't available to those that, by nature of lack of
39 employment or whatever, don't have any coverage at all.

40 DR. McCABE: Michele, I'm going to put you on the spot. Does that come
41 up at all in your advisory role?

42 DR. SCHOONMAKER: Well, I've just been with CRS for only a few
43 months, so I'm not really at liberty to discuss what policies Congress may or may not be
44 considering, but it is a fundamental flaw, if you will, with our health care system and the way
45 health care is delivered in this country. Perhaps if that's something that the committee could

1 contribute to by framing policy issues or making recommendations, I would encourage you to
2 contact your members of Congress or the committees that would have jurisdiction over those
3 issues and presenting an argument.

4 DR. McCABE: Just to remind everyone, though, don't get so excited and
5 call them tonight because you can't do that while you're a special federal employee.

6 Sean?

7 DR. TUNIS: The only thing I would add is it's become a standard part,
8 when I talk about coverage and reimbursement for a new technology, to talk about all of the
9 existing unmet needs for technologies and services that are very high value, very cost-effective
10 or cost saving, that are not part of a benefit package or not particularly highlighted or talked
11 about.

12 I think it's an excellent point to raise here that, even within the realm
13 specifically of genetic services, there may be those that are extremely high value to populations
14 that we tend to forget about, and to take the opportunity to highlight those, rather than entirely
15 being attracted to sort of what's five or 10 years over the horizon in terms of the fanciest stuff
16 for people who have insurance. I think that's an important element to every discussion about
17 new technology. We've got a limited amount of resources to spend in health care and I don't
18 necessarily think that we're attentive enough to the notions of getting the best value for
19 resources.

20 DR. McCABE: Anyone else on the panel want to take a shot at that?

21 (No response.)

22 DR. McCABE: If not, Hunt, and then Cindy.

23 DR. WILLARD: I was struck, and Marc, I think you said it most eloquently
24 by saying you were whining, but four or five of you, the obvious take-home lesson from
25 everything you've presented was we want to be paid more for our laboratory tests or we want to
26 be paid more for our clinical services. Yet I've spent enough time in hospital CEO offices to
27 realize that that's not going to happen anytime soon.

28 So the ray of sunshine here was Dr. Veenstra. So my question to him, and
29 the whiners can chip in as they will --

30 (Laughter.)

31 DR. WILLARD: My question to him is how large a group do you
32 represent? I know of only a few groups like yours nationally who are really trying to address
33 this issue of cost-benefit analysis in order to make an argument to someone that in fact this does
34 eventually pay the system back or even save money in the long run, even though in the short
35 run it's probably much more expensive. So how rare a bird are you?

36 DR. VEENSTRA: An N of 1.

37 (Laughter.)

38 DR. VEENSTRA: No, my perspective is definitely coming more from kind
39 of the pharmaceutical reimbursement area, and I think 10 years ago there was a lot of cost-
40 effectiveness analysis. I think really the methods had been fully developed and there's a lot of
41 research in the area. You didn't see them applied too much. There were guidelines in Australia
42 for requiring cost-effectiveness analyses before reimbursement of tests.

43 Now, we have a situation where in a lot of countries, and in particular in the
44 United Kingdom, there's a National Institute for Clinical Excellence, NICE. A lot of people
45 call it not so nice, but they look at clinical evidence and they also look at cost-effectiveness

1 before they make coverage decisions, and it's having an impact on the way drugs are brought to
2 market and it's starting to impact the pricing, and what I explained to you about this AMCP
3 format, that's really grown over the last few years, and so you're starting to see somewhat of a
4 change of a perspective there.

5 So I would say that there's a strong academic community, but in terms of
6 actually influencing decisions and playing a role in decisions in the real world, I think it's really
7 just starting to happen now.

8 DR. WILLARD: Because I must say, in terms of what the committee might
9 do, that focusing on that issue going forward of how we might take advantage of that body of
10 expertise to examine the future of applying genetic and genomic technology for a larger and
11 larger set of patients is more likely to be well received than simply arguing that a group is
12 underpaid for the services we're already providing, and therefore we'll really be underpaid when
13 we start providing even more. But if we can wrap that around an argument, assuming it works
14 out that way, that this does have a positive cost-benefit ratio, then that would be time well
15 spent.

16 DR. McCABE: Marc, and then Michele in response to that, and then we'll
17 move on to another question.

18 DR. WILLIAMS: Yes, I think that's certainly reasonable. I think, though,
19 it's also fair to look at what is happening in the private sector, and I think Ron's example in
20 Kaiser and to some degree even in our relatively small integrated health care delivery system,
21 there is the recognition, even though we don't really have the capability -- at least our group
22 doesn't, and Kaiser probably does -- to actually internally do those types of studies, I think what
23 you recognize is that some of the relatively low-cost interventions that we do, like taking a
24 family history and doing genetic counseling and providing those services, actually in many
25 cases reduces the number of higher-end technologies that are being utilized. Of the patients
26 that are referred to our cancer susceptibility clinic, only about 1 in 10 actually go forward with
27 a test.

28 So I think there are savings to be accrued to the system by doing things well
29 in the front end, and while I think you're right, particularly when you look at the editorial panel
30 and the fact that there are no family practitioners, internists, or pediatricians that are actually a
31 voting member of that panel, it does tend to understand a little bit about why there's a skew
32 between procedure versus, if you will, cognitive services in this country.

33 But there are cases to be made and within an integrated health care delivery
34 system, I think those cases are being very effectively made. The problem is that that doesn't
35 necessarily translate into the way the care is generally delivered in this country.

36 So I think beyond the academic approach to cost-effectiveness or cost-
37 utility, I think there's some practical experience in well-integrated systems that would
38 demonstrate value and quality as well.

39 DR. McCABE: Michele?

40 DR. SCHOONMAKER: Thank you, and again, these are my own views.

41 I think a problem that precedes whether or not we can use cost-effectiveness
42 information is a problem that we need a better way of collecting data to assess effectiveness.
43 As you heard Sean say, CMS and FDA are both bound by the legislation and the regulations
44 that say that they look at safety and effectiveness or medically necessary and appropriate, and
45 perhaps one thing that the committee could explore would be a better way to or a way to

1 promote the better coordination between those two agencies in the evaluation of new
 2 technologies that would enable you to collect the type of data to make effectiveness
 3 determinations, and then adding the cost may be relatively straightforward or not. I see it as a
 4 way of more efficiently handing the baton from one agency to the other one without stifling the
 5 innovation that's going on in the field.

6 DR. McCABE: That was a windmill that we tilted at under SACGT. It
 7 would require legislative change because of the way the laws, the enabling laws, for the two
 8 organizations -- and the cultures of the organizations. These fundamentals are firmly
 9 embedded. So we had decided not to, while we recognize that it's a fundamental tension
 10 between those agencies, because where one determines that it's safe and effective, the other
 11 determines that it's not valuable enough for reimbursement, so it does create problems and it is
 12 a problem in the system. We could decide if we wanted to take that one on, but it's fairly
 13 deeply rooted, as we learned during those previous deliberations.

14 Cindy?

15 MS. BERRY: One of the frustrating things in trying to change a policy,
 16 particularly in federal programs and Medicare in particular, is the fact that to get a change in
 17 the law, the Congressional Budget Office has to look at the proposed law and determine how
 18 much it's going to cost. Unfortunately, CBO is dealing with narrower budget windows, where
 19 maybe we're dealing with five or 10 years, and maybe some of the research that's out there that
 20 demonstrates real cost-benefit and effectiveness is looking farther out than that.

21 Then on top of that, I think, even though they might not admit it, but you're
 22 cheaper if you're dead than if you're alive according to CBO.

23 So does anyone have any strategies or ideas for how we can combat that?
 24 Because to the extent that some of the changes that we might want to see made require
 25 legislative change, legislation actually passed by Congress, we're going to have to deal with the
 26 Congressional Budget Office and its way of analyzing these things. So it's a perennial
 27 challenge, I think.

28 DR. McCABE: First, I'm going to toss that one to David, and then have you
 29 chime in, Marc, but my understanding at a very superficial level is that's one value of the cost-
 30 utility models because it does put a premium on survival.

31 DR. VEENSTRA: Yes, I think that sums it up pretty well. If you're doing a
 32 cost-utility analysis, you're looking at quality of life and life expectancy as your output. That's
 33 what you're producing. That's what we're all here in the business of, is improving people's lives
 34 and helping them to live longer.

35 So when you're doing that type of analysis, it works well, and it's not best to
 36 let everyone die, but if you are just looking purely at cost and if you don't have any way of
 37 assigning even a monetary, economic value to someone's life, then you're correct. You're
 38 always going to end up that it's cheapest for everyone to pass away.

39 That can be a serious challenge and I think the use of cost-utility analysis is
 40 pretty well entrenched in the academic community, and if there's a need to encourage the use of
 41 it, I think there are a lot of folks that would be willing to help out.

42 DR. McCABE: Marc?

43 DR. WILLIAMS: I think that a lot of the things we're talking about don't
 44 have that long a window. There is evidence emerging in the BRCA group that cost savings
 45 accrue to health plans who cover testing, predispositional testing, to women based on the

1 decisions that they make then to perhaps undergo either hormonally-based therapies, preventive
2 therapies, or surgical preventive therapies, that the cost savings in a large group of insured
3 actually accrues within one to two years. So we're not looking at a 10, 15, 20-year window.

4 I think also with some of the ability to detect susceptibilities to drug
5 reactions, a couple that were mentioned today, and then the other one I would put on the table
6 would be malignant hyperthermia, where we have the ability to identify about 80 percent of the
7 individuals that are susceptible to malignant hyperthermia reactions to anesthetic agents and
8 those can be easily avoided then.

9 The cost there would be immediately recognized as soon as that person
10 either was going to be exposed to that drug or underwent that procedure because then we would
11 be avoiding a medical catastrophe that would have attendant costs associated with it.

12 Again, I think some of the graphs that were presented by Dr. Veenstra
13 clearly showed that at even relatively low prevalence levels, these can be identified and be very
14 cost-effective. So I don't think, even when we're looking at chronic diseases, we're not
15 necessarily looking at a 15 to 20-year payback on some of the investment.

16 DR. McCABE: Yes, David?

17 DR. FEIGAL: Is there any estimate in the genetic testing area or from any
18 of the advocacy groups what percentage of that testing ends up being out of pocket for patients?
19 I remember when the SACGT was meeting, there was some testimony presented. I remember
20 one scenario where they had to pay \$2,500 per family member per test, and it could only be run
21 if they paid for the test because it wasn't covered.

22 We've got the figures in round numbers for drugs. Do we have the same
23 thing for diagnostic tests?

24 DR. McCABE: Andrea?

25 DR. FERREIRA-GONZALEZ: Yes, I don't currently have those numbers
26 and it may be a little difficult to get to those numbers because a portion of the way we track the
27 reimbursement, like at a large academic center, you bill as a global billing, where the patient
28 comes in, sees the physician, and there's radiology, pharmacy, and laboratory practices. So a
29 bill is sent to the third-party payers, but also the division might choose to pay out of pocket the
30 genetic test. So they will be assigned a different account and so forth. So it would be very
31 difficult to track that information.

32 We can try to get that information for the committee for the future. I think
33 it would be very interesting to see that.

34 DR. FEIGAL: Part of what makes this challenging is sort of the increasing
35 practice that you don't have to go to the laboratory to get your test done because the samples
36 can travel to central laboratories, and so there may not be a physician interaction and there may
37 not be an institutional interaction. Some of the examples are actually from academic medical
38 centers, where they might be providing a scarce test for people all over the country, and when
39 you've got that kind of arrangement, I imagine it's more challenging to figure out the billing, but
40 it would be interesting to sort of look at that dynamic.

41 DR. McCABE: Debra, you want to follow up on that? And then Marc.

42 DR. LEONARD: Well, as a laboratory director, that's hard to determine.
43 My laboratory does get testing from across the country and even internationally, and we ask
44 everyone outside of the University of Pennsylvania Health System to pay upfront for the test.
45 So there's this trickle-down effect that we aren't going to eat the cost because the

1 reimbursement is so poor. So therefore, we ask for full payment of our charge, but then
2 downstream, the patient has to work with their third-party payer or the health system has to bill
3 or the health care provider or the genetic counselor, and then they get the poor reimbursement.
4 So we actually prefer to do testing outside of the University of Pennsylvania than for our own
5 patients because the reimbursement is close to 100 percent.

6 DR. McCABE: Marc, is your comment on this point?

7 DR. WILLIAMS: Yes, and I think the point that we don't want to miss here
8 is that looking at it from the laboratory perspective backwards, in addition to the problems
9 noted about that, it's going to miss the other side of the equation, which is those that chose not
10 to undergo testing when they realize that it's going to be out of pocket.

11 As the person that sits down with the patient and works with the insurance
12 companies and tries to get the payment for the testing upfront, I can say that we probably are
13 successful, depending on the test, anywhere from -- well, anywhere from 0 to 100 percent, but
14 for the common ones, the BRCA and the HNPCC, we probably bat about 400 or 500 in terms of
15 getting third-party reimbursement, and then those other individuals are left either to pay out of
16 pocket or we actually have established a fund for those that are really in dire financial need.
17 We can actually subsidize testing in those individuals, thanks to a generous donation.

18 But it's going to have to be a two-pronged approach to try and get a handle
19 on that because I think there really are a lot of people that would like to be tested, but choose
20 not to be tested because of the out-of-pocket expense.

21 DR. McCABE: And certainly, that gets back to the stratification issue that
22 Joan pointed out before.

23 Emily?

24 DR. WINN-DEEN: I guess I want to put the CMS guys on the hot seat for a
25 second here because, despite all these presentations, I still don't get it.

26 If I have a new test and I come out with that, I can put all my stackable
27 codes together and figure out how many PCR reactions and how many probes and what the
28 Medicare default reimbursement might be, but if I had an excellent health economic argument,
29 what I heard is that CMS still wouldn't change the reimbursement. They would only use that to
30 determine if something would be reimbursed.

31 So it sounds to me like despite whatever value a test could bring to the
32 overall health care system, that at least for the next five years until 2009 when lab fees are
33 unfrozen, that we're stuck.

34 DR. McCABE: Don't?

35 MR. THOMPSON: Sure. It's multiple parts to the question, but let me try
36 to address it in pieces.

37 To the extent that this new technology or this new test could be broken
38 down into existing CPT codes, you are correct that it would be frozen until 2009, and the only
39 avenue that CMS has available to it, and we're not quite there yet, is what I spoke about
40 inherent reasonableness, which is our ability to look at a fee and say this is inherently
41 unreasonable and there is a rather long process in the law for changing that payment amount.

42 The reason that's a long process is they were more terrified about us
43 reducing fees than they were increasing them, but it's the same process. The sword cuts both
44 ways.

45 So to the extent we have a new technology covered by existing CPT codes,

1 we're a little bit handcuffed in our ability to change those payment amounts, though I did
2 mention IR, inherent reasonableness, is in the future and one of our first candidates is the
3 HIV/HCV viral loads. That's one avenue, but that is again kind of a brick-by-brick approach.

4 Now, to the extent that you have a new CPT code for this test or there's no
5 way to add the component pieces in the existing codes or we require a new CPT code and that
6 comes into play, now it goes through the CPT process and the code is approved, in that
7 scenario, that's where kind of the 942(b) comes in, the ability that Congress said, well, publish
8 some regulations on how you want to go about doing new lab tests.

9 Again, one might argue that the directions they gave in the statute would
10 mean just take your current process and put it in regulations, but that's why we're going through
11 notice and comment.

12 So it's kind of a little bit of a, like I said, window, a small ray of hope, if you
13 will. To the extent that you get a new CPT code, there is some flexibility in there on how we
14 might set the price.

15 So is that helpful? Probably not.

16 DR. WINN-DEEN: Yes. I guess I'm still concerned that you could have a
17 test that has got a huge amount of economic value and CMS still would not recognize that
18 economic value, no matter what your health economic argument would bring to bear.

19 DR. McCABE: Sean?

20 DR. TUNIS: Yes. You know, the fact that for the most part Medicare
21 doesn't use cost-effectiveness or economic evaluations as part of either coverage or reimburse
22 has been 20 years of intense lobbying by the medical device industry against that, and the
23 reason for that obviously is they'd like to be able to charge high prices for things that don't
24 create a lot of value, but the downside of it is that you can have very high-value things that you
25 don't get fair prices for.

26 I think, and maybe this committee wants to take it on, that there is no
27 sensible way to get good value out of health care resources, whether it's Medicare or elsewhere,
28 without having the ability to do the kind of work that was discussed here today and make it
29 influential in coverage and payment policy.

30 But it's not because -- you know, Congress put those prohibitions in place
31 pretty much under pressure from industry lobbying efforts that have undermined several efforts
32 to make cost-effectiveness a factor in reimbursement policy in Medicare.

33 DR. McCABE: Debra, a comment on this point?

34 DR. LEONARD: Yes, a question. Wouldn't Section 942(b) apply to the
35 new alphanumeric modifiers? So with this whole alphanumeric modifier system that was
36 proposed by the Genetic Testing Work Group that we could do something for genetic testing
37 reimbursement?

38 MR. THOMPSON: That's one of the things we intend on seeking comment
39 on in the new regulation on the 942(b), is when is new new? And that's an open issue.

40 DR. LEONARD: Because this is completely new.

41 MR. THOMPSON: Right. Understood.

42 DR. McCABE: Yes, Andrea?

43 DR. FERREIRA-GONZALEZ: But these new codes are going to modify
44 existing CPT codes.

45 DR. LEONARD: Don't point that out. They're new.

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(Laughter.)

DR. FERREIRA-GONZALEZ: They know that already.

DR. LEONARD: I know that.

DR. FERREIRA-GONZALEZ: But we still bring in new technology or new tests that have clinical necessity and will be effective in treating the patient, but even the modifiers, the only thing it's going to allow is for third-party payers to identify what they're paying for.

Now, the new CPT codes that are going to be coming down the pike, hopefully to address microarray technologies, et cetera, hopefully they will be put in a different pot and seek advice from laboratories, end users, and advice from community associations to see what is the level of reimbursement and so forth.

That's a statement and a question.

MR. THOMPSON: Again, not to be evasive, but I don't like to get in front of my chain of command in terms of the clearance process. We are going to go through notice and comment rulemaking in the issues you just brought up with the modifiers, and without question, the issue of the new CPT code, that does fall into kind of the new technology pot.

That's not to say that if you get new CPT code and we look at it and we see it as cross-walking in a straightforward manner to an existing CPT test, that we might not be in position where, again, we'd be somewhat handcuffed, but granted at least it's in the new code process. The modifier is a little more of an open issue, but at least if you get a new CPT code, it definitely runs through that process where we have a public meeting, we get input, and we decide gap filling versus cross-walk.

DR. McCABE: I just want to make a comment and point out something that was said by Sean, and that is that there might be some benefit to looking at cost-effectiveness for these tests, and also to remind everyone that coverage and reimbursement was in a Category 4, which suggests that it needs more discussion, and that we have both agency public representatives and representatives from private companies here and we've been told that the reason we have this system is because of lobbying by medical device companies, but in fact for testing we have those individuals represented on this committee.

So as we're thinking about our deliberations and how we are positioned to look at recommendations, it seems to me that we have a forum here with the appropriate people sitting around the table for pursuing these discussions.

Chris?

DR. HOOK: Thank you.

I want to just throw a question out to Dr. Veenstra. One of the issues that we're going to be talking about tomorrow is to pursue a bit further the question of pharmacogenomics on a larger scale, and I'm not aware, but I don't know if anyone has informally done the thought experiment along the line of if we look at the JAMA study from '98, 106,000 people per year dying from attributed drug reactions, of which only 5 to 10 percent were probably preventable because of clerical error or physician misprescription and so on.

If the country or if the FDA were to go to the effort of or have the law modified to require now submission for new drug approval require pharmacogenetic information be included, what's sort of the cost-benefit analysis that that might bring?

DR. VEENSTRA: That's a great question. From what I understand in swapping voicemails with Katherine Phillips, I think basically planning for doing a study like

1 that is underway right now. I think we'll be working with the FDA as well as some other folks
2 in trying to get a general ball park idea of what that might look like.

3 It is going to be a bit of a thought experiment because we don't have a lot of
4 these association studies, but just laying out the basic parameters, where do we end up? How
5 many lives are saved in general? Is there any chance that it might make sense? So I think it's a
6 great question and it's something we're going to try to look at.

7 DR. McCABE: Before we go to Joan, Sarah was just showing me on her
8 Blackberry here one of her latest entries is "Gene Testing Families Risk Overheating Updated."
9 Monday, March 1st, at 4:27 p.m., so extremely current, and it's by an AP medical writer,
10 Lauren Neergaard, N-E-E-R-G-A-A-R-D, and basically about Uncle Joe waking up from minor
11 surgery packed in ice, so it was the malignant hyperthermia, which not only has morbidity but
12 also mortality still associated with it. We didn't scroll down to read the whole article. I'll leave
13 that up to you tonight to catch it on CNN, but certainly very timely.

14 Joan?

15 DR. REEDE: Just a point because we had the other earlier discussions
16 about population studies, and one of the concerns I have when we start talking about
17 pharmacogenomics and these types of issues are assumptions that end up being made that the
18 answers that you get from these studies are able to describe or explain variation that may also
19 have, and most probably has, environmental or other factors involved, and real risk. I think
20 here about some of the discussions that I have had with people who make assumptions that
21 many of the health disparities and other things we see can be explained just on the basis of
22 genetic variation.

23 I'm just concerned that as we go down this path of pharmacogenomics and
24 we start looking at cost-benefit and we start getting into these fine numbers, that we leave out
25 the environmental influences and just want to make sure that as a committee we don't fall sway
26 to the idea that genetics by itself is going to answer these types of issues.

27 DR. McCABE: Emily, you want to comment on that, and then we'll go to
28 the panel?

29 DR. WINN-DEEN: I just wanted to also encourage you while you're
30 thinking about this thought experiment to think about the fact that the really severe ADRs, the
31 ones that you would most like to prevent, are the ones where we'll never have enough statistical
32 power, probably, to find out what the underlying genetic lesion, if it was there, was because
33 you're never going to leave something on the market long enough for the right statistical
34 number of people to die to do the study.

35 So it means that that adverse drug reaction benefit to genetics is not in the
36 really severe, severe reactions. It's in the sort of moderate, like the hyperthermia kind of things,
37 but things that induce people to die, I mean, we've seen how many drug withdrawals in the last
38 five years because of unexplained deaths. I mean, it's not ethical to leave those kind of things
39 on the market.

40 DR. VEENSTRA: That's another potential study where you're thinking
41 about what's been the loss to society of having to pull those drugs off the market? All the
42 money that was spent to develop them. They could benefit patients, but maybe because of
43 genetic variation, we've had to pull them off the market. So I think that's a strong belief of mine
44 is that most of the drugs that cause a lot of problems because of pharmacogenomics aren't on
45 the market.

1 DR. WINN-DEEN: Yes. The question is just can you ever actually find
2 out? It's not that I don't believe there might be a genetic source to that adverse drug reaction,
3 but can you design a statistically valid multivariate study to actually find whatever the genetic
4 lesion or combination of six or eight genetic lesions that cause those few individuals to have
5 this very severe reaction? You know, it's a statistics problem.

6 DR. McCABE: It is influencing care. I think I've mentioned before at this
7 committee the fact that we had a threat of a pharmacogenomics lawsuit at UCLA when a child
8 was on an aminoglycoside, gentamicin, and failed the hearing screen when they were leaving
9 the NICU, and the family threatened to sue us, having gone to the Internet and looked up
10 aminoglycoside-induced hearing loss.

11 It turned out the child did not have the mitochondrial mutation, but I was
12 speaking and actually presented this at a forum at UCLA recently, and the head of neonatology
13 pointed out to me -- this was two years ago -- they had stopped using aminoglycosides as one of
14 their first-line antibiotics and had gone to a much broader spectrum, cephalosporin, and now
15 we're seeing the consequence of that that was predictable with a lot more Gram-negative-
16 resistant organisms. So there are all kinds of consequences to these decisions that could be
17 ameliorated by the appropriate testing.

18 Marc, did you have a comment?

19 DR. WILLIAMS: Yes. This is related to something that I maybe read
20 between the lines appropriately or inappropriately about the comments that you were making,
21 and I think it is an important issue because there are certainly a group of individuals that are
22 looking at genetic variation with an agenda to say that, well, a lot of the disparities that we're
23 seeing are really buried in this genomic variation, and of course then that also can sometimes
24 get translated into other issues relating to race, ethnicity, and what have you.

25 I just wanted to let the committee know, if you're not already aware, that a
26 group of population geneticists, particularly Dr. Len Jordy at the University of Utah, have been
27 doing some really outstanding work looking at some genetic variation within certain disorders
28 that we've recognized as occurring within a higher frequency with certain racial groups and
29 have not really found that this is a race-specific variation, that this is actually much broader.

30 So I think that what's actually going to come out of the science as we begin
31 to understand the variation more is that it's actually going to have less of an influence on the
32 types of disparity, particularly racially and ethnically-based disparities, that have previously
33 been looked at. But I think if this is work that you're not familiar with, it would certainly be
34 reasonable to have a presentation on that.

35 DR. McCABE: Thank you.

36 Yes, Joan?

37 DR. REEDE: A follow-up to that. I think the important part here is also
38 perception, and if you look at what the public perceives as what you're going to be able to do
39 with genetic testing or pharmacogenomics, there's this sense by some of the public that this
40 testing is going to tell you the difference based on race/ethnicity or deal with all of the health
41 disparities, and there are many other variables that are there.

42 So I think there's a risk of going down a path that we've been down in the
43 past that is not a healthy one for our country in terms of thinking that the science is going to be
44 able to explain some things that it may not be able to explain.

45 DR. McCABE: Martin?

1 MR. DANNENFELSER: Just a clarification on two points for the folks
2 from CMS. You talked about reimbursement and I guess through Medicare, it sounded like in
3 your presentation, for the genetic testing. Is it only through Medicare or is there any kind of
4 reimbursement through Medicaid for genetic testing?

5 That's one, and there will be a follow-up on that.

6 MR. THOMPSON: Sure. Yes, but in the Medicaid program, again, that's
7 kind of a decentralized program, so individual states are making individual terminations. Some
8 of them look at our fee schedule amounts when they're setting those rates. As I think some of
9 the earlier slides showed, they look at our kind of national limitation amounts in setting those.
10 That's not something we control at CMS. That's kind of more at the individual state level,
11 although many of them do mirror the clinical lab fee schedule.

12 MR. DANNENFELSER: But they have the ability or allow the use, if you
13 will, for a broad range of genetic testing at the state level?

14 MR. THOMPSON: I'm speaking from a payment perspective. I'll leave it
15 to Sean from a coverage perspective.

16 DR. TUNIS: It's pretty much the same situation regarding the medical
17 necessity issue in that it's a decentralized decisionmaking subject to state laws, state
18 policymaking, sometimes more generous than the Medicare national policy. Sometimes they
19 look to the Medicare national policies or they'll look to some of the local policies within the
20 state, but there are no mandates from the Central CMS on what the state Medicaid programs
21 would cover.

22 MR. DANNENFELSER: And with respect to Medicare, are there any
23 eligible, is it people, in terms of Medicare who would be seeking reimbursement for prenatal
24 genetic testing or is it other kinds of genetic testing?

25 DR. TUNIS: It's possible under the -- you know, since it's elderly, disabled,
26 and end-stage renal disease, presumably within the disabled category there are some folks who
27 would be seeking prenatal testing. It's just not particularly common.

28 MR. DANNENFELSER: Thank you.

29 DR. McCABE: Debra, and then Brad.

30 DR. LEONARD: Could I ask for clarification? From the private insurance
31 perspective, it's said that you had to have the test be FDA-approved or cleared in order to be
32 paid, and then you guys said that you would also consider laboratory-developed tests for
33 payment. So can you clarify whether tests are only paid for if they're FDA-cleared?

34 DR. McCABE: Michele?

35 DR. SCHOONMAKER: I'm sorry. That was FDA-approved or those that
36 conform to the CLIA requirements. So if they're performed in a CLIA-certified laboratory, then
37 that would be the regulatory approval that would be pertinent.

38 DR. LEONARD: So FDA approval is not required.

39 DR. SCHOONMAKER: Not for tests. Right.

40 DR. LEONARD: As long as they're performed in a CLIA-certified
41 laboratory.

42 DR. SCHOONMAKER: Right.

43 DR. McCABE: Marc?

44 DR. WILLIAMS: It ain't that simple. The problem is that each private
45 insurer can make their own decision and they can decide that they will only pay for FDA-

1 approved laboratory tests.

2 So while many insurers will look to CMS regarding coverage and payment,
3 they're certainly not obliged to follow that, and there are no other national things that they need
4 to follow, and so basically they're all on their own, which leads to the frustrating situation that I
5 know you've experienced, because I know we've experienced it, which is you have to go insurer
6 by insurer and basically fight that battle with every single one of them. There's no shortcut to
7 be able to do that.

8 Again, the FDA approval has to do with a situation where you have to have
9 something that says somebody's looked at this, and that's sort of been the standard, but it just
10 doesn't fit the paradigm of these individually-developed tests.

11 So we've actually referred insurers to the American College of Medical
12 Genetics testing, voluntary testing, and saying this is really the standard that the industry is
13 using and so this is really what you should be asking the laboratories about because the FDA, at
14 this point at least, does not have the full jurisdiction over these.

15 DR. LEONARD: Well, with the coding system the way it is, also I don't
16 know how payers know, since all genetic tests use the same codes, what would be FDA-
17 approved and what wouldn't and what would be laboratory-developed and what the testing was
18 even for.

19 DR. WILLIAMS: You've got that right.

20 DR. McCABE: Brad?

21 MR. MARGUS: Just in response to Joan, I wanted to mention that my
22 company is involved in doing massive association studies with large numbers of cases and
23 controls and what we think are sufficiently powered enough to find things that are real, and
24 while a lot of scientists publish those things all the time, I think most geneticists are now
25 arriving at the conclusion that you really need to replicate in other populations and other
26 environments. So there is pretty much an effort in place to not jump to conclusions that you
27 definitely know it's genetic until you actually can prove it in different environments and
28 replicate it. So you should feel better about that.

29 I really appreciated the tutorial today for all this. I guess I have a question
30 about if you came up with a new association, an anomalous association that has tremendous
31 value, even though I understand CMS may not care, and you go about making a CPT for it, my
32 first question is how long does it typically take to get a new CPT?

33 Then the second question is this subject of cross-walking and gap filling.
34 So if the new test, the content for that test, the loci that you're interrogating are new but the
35 approach is still a PCR-based assay that just has different primers or something like that, do
36 you then conclude that it's cross-walking and you actually can easily find and you already have
37 an established ancient rate that you apply to it or does the fact that it's a novel test give it a
38 chance for a new rate?

39 DR. McCABE: Don't

40 MR. THOMPSON: Sure. The approach that we take, it's on a case-by-case
41 basis. So during that kind of public meeting process, we kind of rely on the industry and
42 clinicians. Any member of the public can come in and give us recommendations, and based on
43 that information we have, we kind of huddle with our clinical staff and our clinical folks and
44 our contractor staff, and we look at that case by case. So there's no blanket rule for when you
45 cross-walk and when you gap fill. It would be dependent on the individual situation and the

1 public input.

2 Then your first question, I'm sorry, was?

3 MR. MARGUS: How long for a CPT typically?

4 MR. THOMPSON: Rapid is not a word that comes to mind.

5 (Laughter.)

6 MR. THOMPSON: It can be a multi-year process. Unfortunately, or
7 fortunately, depending on how you want to look at it, we have kind of relied on the AMA and
8 they have kind of formalized CPT process that they go through. It's a deliberative process, so
9 anybody can request a code, but it is, again, not rapid. It can take a year or two years, and
10 sometimes they can table the code and come back and they may seek additional information.
11 So it can be rather involved.

12 MR. MARGUS: So what repercussions or impact do you think that has, the
13 time it takes? Does it hurt things or not too much?

14 MR. THOMPSON: A loaded question, but if you're asking does the fact
15 that you can't get a code or you don't have a code in a rapid fashion for a new technology have
16 an impact on access, going back again to some of the earlier presentations, there are kind of
17 miscellaneous codes that you can go under, but the problem is there's a certain administrative
18 burden associated with going down that road. If you have your own code, it's in the system. It's
19 a much more rapid administrative fashion. You can have a public discourse on the price at a
20 national level, whereas when you go with some of the more miscellaneous codes, you may be
21 able to get access, but there might be an administrative burden associated with that.

22 I'm sure, given the reaction from both sides, I think, that we have some
23 thoughts on the CPT process.

24 DR. McCABE: If this is a comment on this, and then Joan and Hunt, and
25 then we're going to wrap up.

26 Andrea?

27 DR. FERREIRA-GONZALEZ: I want to bring an additional issue. It
28 seems to me that a way to look at changes in the reimbursement for Medicare is through the
29 inherent reasonableness. My question is when can we expect this process to take effect and
30 maybe if there is anything that this committee can do to provide reinforcement to the Secretary
31 to move this process faster?

32 MR. THOMPSON: Sure. As I mentioned, we were under a congressional
33 moratorium with respect to inherent reasonableness for many years. Again, I think out of fear
34 that we were going to reduce prices. That moratorium has recently been lifted and in the wake
35 of that, we are now struggling with coming up with instructions for how that process is going to
36 work because we want to ensure that we have a kind of fair and equitable process because it
37 will be used in both directions. You know, prices can come down and prices can go up under
38 it, and it's going to be the same process either way and we make sure, given the additional
39 congressional input that we received on inherent reasonableness, that we follow that. So our
40 hope is that this year that we will be able to issue those final instructions on inherent
41 reasonableness and then begin the process for changing those payment amounts.

42 Again, both up and down, and this is kind of focused on genetic testing, but
43 in kind of looking philosophically at the clinical lab fee schedule globally, we have a number of
44 tests that one could argue, and some have argued, are in fact overvalued. For example, if one
45 looks at urinalysis CBC, some of which are kind of the bread and butter of some labs, and even

1 some, I would argue, at VCU, to the extent we're overpaying for those and we decide to go after
2 some of those high-volume tests, which may represent 50 to 60 percent of the clinical lab fee
3 schedule, the hue and cry if we don't get the process right will be loud. So we've got to make
4 sure we get it right for both the increases and the decreases.

5 DR. McCABE: And a follow-up on that, is there anything this committee
6 could do?

7 MR. THOMPSON: At this point, I can't tell you the internal pressure to get
8 those turned around, so I can't imagine adding another voice would cause the process to be any
9 more rapid, but again, the committee may feel otherwise.

10 DR. LEONARD: Can I clarify something on Brad's comment?

11 DR. McCABE: Yes.

12 DR. LEONARD: Which is that the understanding is that you do not add
13 new CPT codes for every single genetic locus. Otherwise, we could end up with 25,000 times
14 however many mutations there are per gene.

15 MR. MARGUS: So it has to be a new technology for even analyzing.

16 DR. LEONARD: It would be technology-based. Right now, a laboratory
17 implements a new test that classically would have gotten a new CPT code using these -- isolate
18 DNA, do a PCR, run a gel codes, and so we don't go through the new CPT code process.

19 MR. MARGUS: But if it were just a new set of SNPs tomorrow for looking
20 at something else, they already have a CPT for it, they'll apply that rate, and then it's five bucks
21 whether it's worth \$50,000 in savings --

22 DR. LEONARD: Yes. You've got it. So all the new genetic tests that are
23 going to be coming out are constrained by this CPT coding reimbursement process.

24 MR. MARGUS: Everybody just closes their eyes and says wait until 2009.

25 DR. LEONARD: And it's not clear that in 2009 it will be freed up. It could
26 still remain frozen, right?

27 MR. MARGUS: So one question is, Ms. Lab Lady, aren't there any drugs,
28 aren't there any tests --

29 PARTICIPANT: She's Dr. Lab Lady.

30 (Laughter.)

31 MR. MARGUS: Sorry. Dr. Lab Lady, aren't there any tests over the last
32 five years or whatever since the prices were locked in where a technology has helped to reduce
33 the cost and now you're making buckets?

34 DR. LEONARD: No. I can emphatically answer that no. There probably
35 are technologies out there that would reduce costs, but because of the health care finance
36 system, we have no capital equipment budget, so we can't buy robots for automated nucleic acid
37 extractions and 96-capillary electrophoresis instruments and those kinds of things. So
38 technologies may be out there, but the hospital systems in academic health centers where this
39 testing is performed generally don't have capital equipment budgets to be able to be proactive
40 and think about reducing costs in these ways.

41 DR. McCABE: For example, when they got the capillary system in our
42 sequencing lab, then they were able to decommission the old gel-based systems. We gave the
43 one that we had contributed years ago to the sequencing corps, we gave that to our orphan
44 disease testing lab, so they could start doing sequencing. So they're using ancient technology,
45 but it's because it was free, and that's the nature of the capital improvements in our academic

1 health centers.

2 Joan, I'll give you the last word for the day.

3 DR. REEDE: Thank you.

4 Notwithstanding the research and the work that Marc is mentioning is
5 being done or the work that Brad is mentioning with regard to his company, I think that we
6 would be remiss if we did not also take into consideration the fact that not everybody is of the
7 same opinion or sway that we are. So if you look in the literature, you can see articles written
8 by physicians talking about "I am a racially profiling physician," using our genetic information
9 as their justification for racial profiling.

10 So I think, as we talk about our prioritization and we talk about issues such
11 as education and we talk about issues such as making the public aware and moving forward,
12 that we have to be cognizant of the fact that not everyone is going to use this information in a
13 way that we may think is appropriate.

14 DR. McCABE: Thank you very much. That's a very good way to end the
15 evening. Thank you everyone for putting in a long day today.

16 Let me remind you that you do not fill out your second straw vote ballot
17 tonight. We will hold that for tomorrow.

18 I want to thank our presenters again for your time and your participation. It
19 was extremely helpful.

20 I want to remind those who are joining us for dinner -- committee members,
21 ex officios, presenters -- please meet in the lobby at 6:40 p.m. and we will then take cabs to the
22 restaurant. Our reservation is at 7:00 p.m.

23 Just to remind you, because we have two busy days, we are starting earlier
24 than is our custom tomorrow. So we will be starting at 8:00 a.m. tomorrow, and that will be
25 with public comment, so I would ask all of the committee members to be here to be respectful
26 of the public comment session.

27 Thank you.

28 (Whereupon, at 5:35 p.m., the meeting was recessed, to reconvene at 8:00
29 a.m. on Tuesday, March 2, 2004.)

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