

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

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

Fourth Meeting

Monday,  
June 14, 2004

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

## IN ATTENDANCE:

Chair

Edward R.B. McCabe, M.D., Ph.D.  
Professor and Executive 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.

Update on Genetic Nondiscrimination  
Legislation and the Activities of the  
Coalition on Genetic Fairness

Joann Boughman, Ph.D.  
Executive Vice President  
American Society of Human Genetics

## Discussion and Next Steps

Presentation on Information Gathered on  
Efforts in Genetics Education and Training

Joan Y. Reede, M.D., M.P.H., M.S.  
Chair, Education Task Force

Roundtable with Selected Organizations on  
Their Efforts in Genetics Education and Training

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

## Discussion of Draft Education Resolution

Report on the Advisory Committee on Heritable  
Disorders and Genetic Diseases in Newborns  
and Children

Reed V. Tuckson, M.D.  
Liaison Member, ACHD

## Discussion

C O N T E N T S

## Public Comment

Kathleen Rand Reed, B.A., M.A.A.  
The Rand Reed Group

Andrew Fawcett  
American Board of Genetic Counseling

Barbara Handelin, Ph.D.  
Handelin Associates

Overview of Draft Coverage and  
Reimbursement Report

Cynthia E. Berry, J.D.

Discussion of Draft Report and Development  
of Recommendations



P R O C E E D I N G S

(8:39 a.m.)

1  
2 DR. McCABE: Well, good morning, everyone, and welcome to the fourth  
3 meeting of the Secretary's Advisory Committee on Genetics, Health, and Society. The public  
4 was made aware of this meeting through notices in the Federal Register, as well as  
5 announcements on the SACGHS website and listserv.

6 First, I want to begin by taking note of some changes in our membership.  
7 We are very pleased to welcome a new ex officio member, Dr. Howard Zucker, who is the  
8 Deputy Assistant Secretary for Health and is representing the Assistant Secretary for Health in  
9 the Department of Health and Human Services.

10 Dr. Zucker trained in pediatrics and has held faculty appointments at Yale  
11 University School of Medicine, Columbia University College of Physicians and Surgeons,  
12 Cornell University Wyle College of Medicine, and the National Institutes of Health. Dr.  
13 Zucker also served as a White House fellow and worked at the Center for Space Research at  
14 MIT. Welcome.

15 Dr. Steven Gutman, Director of the Office for In Vitro Diagnostics  
16 Evaluation and Safety, has been named as the new ex officio for FDA, replacing David Feigal,  
17 who recently accepted a position at NDA Partners, a product development consultancy to the  
18 biopharmaceutical and medical device industries.

19 In addition, we welcome Joe Hackett from the Food and Drug  
20 Administration, filling in for Dr. Gutman this morning, and Dr. Hratch Semerjian, Acting  
21 Director for the National Institute of Standards and Technology, will be representing the  
22 Department of Commerce for Arden Bement, who is currently Acting Director of the National  
23 Science Foundation.

24 Also, Dr. Steve Phurrough will be substituting for Sean Tunis today. Dr.  
25 Phurrough is Director of the Coverage Analysis Group at the Centers for Medicare and  
26 Medicaid Services. Judy Yost will be here tomorrow representing CMS.

27 Dr. Sherrie Hans will represent the Department of Veteran Affairs for Ellen  
28 Fox. Ms. Amy Turner is here representing the Department of Labor for Tom Alexander.

29 Welcome to everyone.

30 I also want to explain a couple of absences in the membership. Christopher  
31 Hook will be joining us later today. Hunt Willard will join us tomorrow, and may be able to  
32 participate for part of today's meeting by phone. Reed Tuckson will be joining us by phone this  
33 morning for the remainder of the day.

34 Eric Lander is also not present. Last week, with great regret, Eric resigned  
35 from the committee, due to extensive commitments and responsibilities as Director of the new  
36 Brod Institute of MIT and Harvard that prevents him from having a more active role in the  
37 committee's activities. Eric told me that he very much wishes to stay involved in the  
38 committee's work, and I promised him that he would become a distinguished member emeritus  
39 and that we would feel free to tap his expertise as needed. The Secretary will be appointing a  
40 new member to take Eric's place in the very near future.

41 Before I review today's agenda, I would like to remind everyone that at the  
42 end of the March meeting, we set some extremely ambitious goals in preparation for this  
43 meeting. We planned a number of short-term actions, including sending a second letter to the  
44 Secretary in support of federal genetic nondiscrimination legislation.

45 We decided that education and training of health professionals in genetics

1 was a high priority issue, and that after additional fact-finding on genetics education efforts, we  
2 would be equipped to consider a resolution regarding these efforts at this meeting.

3 An Education Task Force chaired by Joan Reede has been hard at work  
4 gathering data and drafting a resolution for our consideration today. The committee also  
5 decided to prepare a report describing SACGHS' mandate and priorities, and providing  
6 background information policy considerations on priority issues.

7 Sarah Carr and her staff, with the assistance of Emily Winn-Deen, prepared  
8 a draft vision report for our consideration. In March, we also identified high priority issues that  
9 require more in-depth study. Coverage and reimbursement of genetic technologies and services  
10 was identified as the first topic on which we would focus our efforts.

11 Staff, with the assistance of Cindy Berry, carried out additional data and  
12 information gathering, and prepared a draft report for our consideration. This afternoon, we  
13 will be discussing the draft, and developing recommendations for the Secretary.

14 Three other issues were also identified as warranting in-depth study. Large  
15 population studies, pharmacogenomics, and direct-to-consumer marketing. Our in-depth work  
16 on these three issues will begin at future meetings. With regard to the direct-to-consumer  
17 marketing issue, we also decided to take a short-term step by drafting a resolution expressing  
18 concern about the proliferation of direct-to-consumer advertising of genetic tests.

19 A task force chaired by Chris Hook accomplished that short-term goal, and  
20 we will be considering the draft resolution tomorrow. For this meeting, we hope to finalize the  
21 two resolutions on education, direct-to-consumer marketing, and the report on our priority-  
22 setting process. Given the length and complexity of the coverage and reimbursement report,  
23 and the work that needs to be done to develop consensus recommendations, Cindy and I expect  
24 that we will want to continue refining the report and gathering additional public comment after  
25 this meeting.

26 We will begin the meeting with an update on the status of federal genetic  
27 nondiscrimination legislation by Dr. Joann Boughman. Following our discussions on genetic  
28 nondiscrimination, we will engage in a roundtable discussion on educational issues with  
29 involved professional organizations, and consider the draft resolution on education. The  
30 afternoon is dedicated to discussions of coverage and reimbursement of genetic technologies  
31 and services. Tomorrow we will hear a presentation from Dr. Muin Houry about CDC's  
32 public health approaches to genomics, and deliberate on the vision report and  
33 direct-to-consumer marketing resolution.

34 Finally, we have allowed time to discuss the other issues we prioritized at  
35 the March meeting, and identify next steps. Let me also point out that we have public comment  
36 sessions scheduled for both days. We will hear comments immediately following lunch today,  
37 and tomorrow morning. Thus far, five individuals have registered to provide testimony. If  
38 there are others who wish to give public comments today or tomorrow, please sign up at the  
39 registration desk.

40 We have received a considerable number of written public comments on the  
41 draft documents as well, including requests that we provide additional time for the public to  
42 comment. The comments can be found in your table folders. As our deliberations on the draft  
43 documents proceed throughout the next few days, we will take careful account of the comments  
44 on each document.

45 At this time, we'll hear from Sarah Carr regarding conflicts of interest.

1 MS. CARR: Good morning. Being a member of this committee makes you  
2 a special government employee, and thereby subject to rules of conduct that apply to  
3 government employees. The rules and regulations are explained in a report called "Standards  
4 of Ethical Conduct For Employees of the Executive Branch." You each received a copy of this  
5 document when you were appointed to the committee, and I'm going to review one of the rules  
6 in that document.

7 Before every meeting, you provide us with information about your personal,  
8 professional, and financial interests, information that we use to determine whether you have  
9 any real, potential, or apparent conflicts of interest that could compromise your ability to be  
10 objective in giving advice during committee meetings.

11 While we waive conflicts of interest for general matters because we believe  
12 your ability to be objective will not be affected by your interests in such matters, we also rely to  
13 a great degree on you to be attentive during our meetings, to the possibility that an issue will  
14 arise that could affect, or appear to affect, your interests in a specific way.

15 In addition, we have provided each of you with a list of your financial  
16 interests and covered relationships that would pose a conflict for you if they became a focal  
17 point of committee deliberations. If this happens, we ask you to recuse yourself from the  
18 discussion, and leave the room.

19 Let me add a sincere thank you to all of you for being so attentive to these  
20 rules. We know that the reporting process is time consuming, and we very much appreciate  
21 how conscientious all of you are about fulfilling your duties.

22 Thank you.

23 DR. McCABE: Thank you, Sarah.

24 Now we're going to hear an update on genetic nondiscrimination legislation,  
25 and the activities of the Coalition on Genetic Fairness from Joann Boughman. Dr. Boughman  
26 is Executive Vice President of the American Society of Human Genetics.

27 Protection against genetic discrimination is this committee's top priority, as  
28 we determined in March, and by previous correspondence with the Secretary. We were  
29 encouraged when Senate Bill S.1053 unanimously passed in the Senate last October, 95 to 0.  
30 Following the March meeting, we wrote a second letter to the Secretary urging that continued  
31 pressure be applied to facilitate passage of this bill in the House.

32 Dr. Joann Boughman is here this morning to provide an update on the status  
33 of S.1053 in the House, and the related activities of the Coalition on Genetic Fairness. Dr.  
34 Boughman?

35 DR. BOUGHMAN: Thank you very much.

36 Each of you in the table folders, and outside, have the list of slides, and  
37 we'll go through these slides very quickly, and then I think just get down to conversation and a  
38 couple of ideas that people might have.

39 Don't mistake the bad news slide being blank for the fact that there is no bad  
40 news. The bad news is that absolutely nothing has happened in any real kind of way. That  
41 doesn't mean there has not been activity, but there has been no real noticeable movement  
42 forward.

43 The good news since the March meeting, is that the session isn't over yet.  
44 We still have some time, and we will continue working folks on the Hill in every way that we  
45 can. The other part of the good news is that the members will be going back to their home

1 districts, and we may be able to call on people from home to contact and discuss issues with  
2 their members.

3 As Dr. McCabe just pointed out, 1053 passed the Senate last fall, 95 to 0.  
4 There were three bills that were brought over to the House, 1910 and 3636 actually formally  
5 introduced S.1053, not yet having been formally introduced. Once introduced, HR 1910 was  
6 sent to two committees, and HR 3636 sent to one committee. Now, there are appropriations  
7 committees that do eventually see these bills on the House side, but the committees listed on  
8 this slide are the committees of real action and deliberation for this kind of bill that involves  
9 both work force, and insurance issues.

10 Our plans, or our hopes, have been to in fact get 1053 to committee, and  
11 then to the floor, or around the committees and directly to the floor. Another opportunity might  
12 have been to get 1019 through some committees so that 1053 could end up, or the wording in  
13 1053, the concepts in 1053, could be the goal of compromise. We continue to believe that it is  
14 not useful to have 3636 on the agenda. If you remember, House Bill 3636 is very much a shell  
15 bill, and this is not really a Christmas tree bill as it is referred to on the Hill, but this is just a  
16 skeletal framework so that as it would go through the process, members would get more and  
17 more wording on it, and it would be more and more complicated to in fact negotiate this  
18 through the process.

19 We have had several meetings by advocates. We have had multiple  
20 meetings with the Chamber of Commerce, and the Chamber of Commerce has not been terribly  
21 vocal on this in a negative way. They have made some statements about some definitions in the  
22 wording.

23 We were very pleased, for example, to see a wonderful editorial in the  
24 Washington Post by the Insurance Association that said this genetic nondiscrimination stuff is  
25 out there. We don't believe that the insurance companies are discriminating, but we don't have  
26 a problem with the Congress telling us not to, because we think we don't do it anyway.

27 So in fact, they basically said in a very positive way, go ahead and pass the  
28 bill, we'll deal with the details in some sort of way. We thought that might be helpful and make  
29 some movement, but it has not. In the meantime, many of us continue with meetings on the  
30 Hill, and with members of a variety of agencies. These meetings are now even broadening.  
31 We are not just meeting with staff members of key committee members, we are meeting with  
32 staff members, and with members of Congress who are good friends of the staff members, and  
33 point blank asking them to nudge their friends who are in the right positions to get some of  
34 these things going.

35 We also continue to work on the leadership in the same kind of way, every  
36 opportunity any of us has to make that comment to any of the House leadership or members  
37 who have influence with the House leadership. Let me give you the kind of example beyond  
38 the formal meetings that we have.

39 I had been at a couple of events where Representative Chris Van Hollen  
40 from the District here in Maryland that represents a significant number of scientists, and  
41 FASEB is in his district as well. My conversation centered around our frustration on genetic  
42 nondiscrimination. He agreed with me, and basically I said, let's just not get frustrated  
43 anymore, it is time to get angry, it is time to really get upset about this and move forward. So  
44 those are the kinds of changes that we are trying to make with our friends that may have  
45 meaningful positions.

1           Since the March meeting, we have had a letter from distinguished scientists  
2 sent to the leadership. There have been a series of blast emails that have gone out via CapWiz  
3 and some other venues, to allow letters or emails to go to members. For example, I know from  
4 the American Society of Human Genetics, when we sent out our blast email encouraging all of  
5 our members to email their members, there were 1,100 emails that were received within about a  
6 96-hour, or 92-hour period of time.

7           So in fact, we've had some of these waves of activity, and those were only  
8 the ones that I could count, and other organizations had some of those activities as well. When  
9 1,000 or 2,000 emails show up on the Hill in a two or three day period, what basically happens  
10 is the staff members must take notice of this. When that level of activity happens, they must  
11 report that to their Congressman, and then the discussion goes on in the halls, which may be  
12 another way.

13           We have also prepared a one-pager of information from geneticists that we  
14 have given to the Coalition for Genetic Fairness. We are still working on some more good  
15 stories to get out there as well. We are also asking for more follow-up contacts by individual  
16 constituents.

17           Additional strategies that we would encourage everybody in this room to  
18 put forward are additional personal discussions with members. Maybe we'll just be able to  
19 wear them down. If we can't get them excited about it, maybe we can at least wear them down.  
20 I know that you can hear the frustration in my voice on this.

21           We are going to be continuously requesting personal contact by  
22 constituents, especially when the members are at home during breaks, and encourage strategic  
23 contact between members and leadership. What we have got to do is keep the Genetic  
24 Information Nondiscrimination Act on the agenda. As you all are keenly aware, there are many  
25 other things on the agenda, and the strategy that we just have to keep putting forward is that  
26 this issue, while maybe not as big, or as front page as many of the international, or even other  
27 national events, it is still the right thing to do, and this is something that they can do for the  
28 American public.

29           Hopefully by coming today and venting some of my frustration, at least  
30 among friends and allies, we can state a renewed commitment and gain encouragement from  
31 each other to try and move this forward. At that point, I'll be happy to answer questions. There  
32 are other members in the audience who have been at some of these meetings and have met with  
33 some of the members of Congress.

34           I would ask if any of them have specific comments, just reiterate the fact  
35 that while on the one hand, it is appropriate for us to continuously recognize that this is not the  
36 most important item on the agenda, and that we do have a special interest in this, and it is not  
37 self-interest. It really is a bill for the people out there, and to pass this bill, everybody could go  
38 home with a win.

39           DR. McCABE: Thank you very much, Dr. Boughman, and if you could  
40 join us at the table perhaps for this discussion.

41           While you are doing that, I'll just remind everyone that government  
42 employees and special government employees, and all of us on the committee, while we are  
43 acting for the committee, are special government employees, so we are precluded from  
44 lobbying. Therefore, it is just important to note that this is an update from ASHG, and  
45 represents the views of ASHG and the coalition. Our discussion here is really on how we can

1 provide advice to the Department of Health and Human Services and the Secretary.

2 DR. BOUGHMAN: Dr. McCabe?

3 DR. McCABE: Yes?

4 DR. BOUGHMAN: I would say that I am well aware of that, and hopefully  
5 people understood that this was the activity of an organization here in town, and the update on  
6 those activities, and recognize that we all wear a variety of hats, and that information and  
7 education to ourselves and to others that we work with about the importance of individual  
8 contact.

9 I think one of the roles that this committee can continuously have is  
10 educating individuals about the situation, just where things are in fact.

11 DR. McCABE: I appreciate that very much. We were just clarifying that  
12 the "us" that you were using was referring to the actions of the ASHG and the coalition, not the  
13 actions of this committee. We discussed the possibility in March that further action by our  
14 committee may be required in the event that legislation is not passed in this Congress, and I  
15 hate to be pessimistic by stating that, but I think it is reality.

16 As a reminder, we discussed organizing briefings from individuals who  
17 have faced genetic discrimination, who paid out of pocket to keep their genetic information out  
18 of their medical records, and who chose to forego treatment due to fear of genetic  
19 discrimination.

20 I think it is important to remind ourselves that those were activities we  
21 considered, and that we might consider for the future, should this legislation not pass. We now  
22 have some time to discuss how we should proceed. I would take questions or comments from  
23 the committee for discussion, and particularly since we have Dr. Boughman at the table for her  
24 comment.

25 Dr. Winn-Deen?

26 DR. WINN-DEEN: Yes. I just wanted to ask if you had had any success in  
27 gathering up some of these patient stories. I know you had mentioned at your last presentation  
28 for the committee that you were going to be working on that, and I think that would be  
29 extremely helpful, at whatever point you have those.

30 DR. BOUGHMAN: Yes. In fact, and I'm sorry, I failed to put this on, and  
31 it was a very important event. The Genetic Alliance is a very active member of the Coalition of  
32 Genetic Fairness, and it had a press briefing on the Hill. In fact, there was a very impressive  
33 woman who told of her case. At that time, we were reenergized to gather some cases.

34 We have not created other forums for the release of that information, but  
35 there has been some volleying back and forth. Coalitions of organizations, and coalitions of  
36 coalitions tend to be a little bit slower in getting some things done than we might like  
37 sometimes. But in fact, we have also worked at getting some of these cases, encouraging them  
38 to talk directly as constituents to talk to their congressional representatives, too.

39 I mean, this woman and her two children who were carriers and denied  
40 insurance, made a very impressive presentation. It clearly indicated what the challenge was  
41 before us.

42 DR. McCABE: And we know also of cases where individuals have lost  
43 employment because, presumably at least in one case that I know of, because of concerns  
44 regarding insurance. I know that case appeared before the EEOC. I don't know if you'd wish to  
45 comment, Paul.

1 MR. MILLER: No, I think you articulated the issue.

2 DR. McCABE: Other comments? I would remind everyone that the first  
3 letter of the Secretary's Advisory Committee on Genetic Testing, wrote to the Secretary was  
4 about that, this issue, that our first letter, and the first letter of SACGT to the new  
5 administration, to the current administration, was about this issue.

6 Our first letter to the Secretary of the Secretary's Advisory Committee on  
7 Genetics, Health, and Society was on this issue, and we followed it up with another letter. So  
8 clearly this has been a top priority for these two committees, and I would guess continues to be.  
9 Yes?

10 MR. LESHAN: I'm Tim Leshan, sitting in for Dr. Collins today, with the  
11 Genome Institute, representing the NIH. I just feel like I'd be remiss if I didn't say that Dr.  
12 Collins has been providing technical assistance and working on this issue, both within the  
13 administration, and in responding to requests from folks on Capital Hill about this issue, and  
14 really thanks the coalition for its efforts in this area. NIH feels it is very important that we see  
15 if we can get this legislation passed, as you know. We're very encouraged by some of the press  
16 activity this year, but also concerned that we haven't seen some action, but still hopeful that we  
17 may in the future, and we'll continue to provide assistance and work on the issue as much as  
18 possible.

19 DR. McCABE: Tim, thank you for introducing yourself. I apologize for  
20 not recognizing you in my opening remarks. You are representing Alan Guttmacher, who will  
21 be sitting in for Francis Collins when he arrives.

22 Also, I apologize to Suzanne Feetham for not recognizing you sitting in for  
23 Sam Shekar.

24 Yes, Joann?

25 DR. BOUGHMAN: I also just wanted to remind the members of the  
26 committee that the situation that we're in is that there has been written documentation, the  
27 White House, the administration support of 1053, and it is actually in writing. If 1053 came to  
28 the desk of the President, he would sign it, basically, is the wording in this SAP, as it is referred  
29 to.

30 So that any comments that would be made, or could be made to the  
31 Secretary, would be made in a context of already having documentation from the Executive  
32 Branch in support of the legislation that has passed the Senate.

33 DR. McCABE: Thank you.

34 Other comments? Yes, Emily?

35 DR. WINN-DEEN: Are there some specific issues? Because the last time  
36 we discussed this, it seemed like the issue was, is this really an issue. Have we gotten past  
37 that? Is it just a prioritization versus other more pressing congressional matters at this point?  
38 Or what is the barrier that we need to overcome?

39 DR. BOUGHMAN: I think it is both height and width of the barrier. There  
40 are just so many very big issues going on, budgetary issues being one of them, along with  
41 everything else that is going on, that it is difficult to get the more focused issues onto a series of  
42 agendas. As you are well aware, several things that have happened, in fact, Congress spent  
43 almost all of its time last week on the issues and commentary on former President Reagan. So  
44 what issues were going to be debated during the week last week, slid another week. So that the  
45 challenge is both height and width, I think.

1 DR. McCABE: Paul?

2 MR. MILLER: I would just add that it is always in the context of these  
3 discussions, as I have heard and participated in them, it is always useful to reinforce, and to  
4 have more information about the reality of this problem, about the concerns, and to develop the  
5 issue of the concerns of real people out there, and the impact on the research and science. It is  
6 not the kind of issue that is sort of addressed, and then move onto something else. I think that  
7 that is constantly an issue and concern in the background underlying a lot of these discussions.

8 DR. McCABE: Debra?

9 DR. LEONARD: So beyond writing letters, which we've done now twice to  
10 the Secretary, and we clearly have the Executive Branch's support, what are specific steps that  
11 this committee could take to assist with getting this through the House?

12 DR. BOUGHMAN: My hope would be, and we will utilize the fact that  
13 this issue was on the agenda of this, a federally appointed committee, to look at these issues,  
14 that this was the top priority and the first topic of discussion. That fact will allow us, and  
15 anybody here in the room, to comment on the importance of this issue to the committee,  
16 responding in part to what Paul said.

17 It doesn't enumerate individual issues. But in any way that we can utilize  
18 this support to bring forward more individuals who need to be supported to go see their  
19 Congressman or whatever, and we will be writing additional letters to the leadership and  
20 Congress once again, to try and do this.

21 But we are using just about every venue that we know to use at this point, I  
22 must say. I don't know that there are additional specific actions that we could do at this point.

23 DR. McCABE: Well, I'd just remind everyone that we have discussed the  
24 possibility of having briefings where we would have individuals appear before us who had been  
25 the subject of genetic discrimination. I would ask if that is something that the committee would  
26 like to consider for our October meeting, should the House bill not pass by that time. Again, I  
27 don't wish to be pessimistic, but I think it is important for us to plan if the actions of the  
28 coalition and others were successful, then there would be no need for that, and we could change  
29 the plans for that briefing at that time. Is that something that the committee would wish to  
30 consider? Debra?

31 DR. LEONARD: I think that we should do that. I don't know, five or 10  
32 minutes for each person with a significant number, not just two or three people, so we can have  
33 an impact.

34 DR. McCABE: Okay. Is that something, Sarah, that the staff could work  
35 on for October?

36 MS. CARR: We can certainly try to do that. I guess I would also, if it is  
37 appropriate, to reach out to the coalition, because you've done so much of this already. If there  
38 is a way we can augment that work, or complement it in some way by bringing it to the  
39 attention of this committee, and thereby to the Secretary, we'd certainly want to, and would do  
40 whatever we can.

41 DR. McCABE: Is there anyone on the committee, among the members or  
42 the ad hoc representatives, who would disagree with that plan for October?

43 (No response.)

44 DR. McCABE: Okay. Seeing no one wish to comment, then I'll assume  
45 that we will move forward and begin planning for that at the end of this meeting.



1 MS. CARR: Maybe it would be helpful if some of the members, we could  
2 form a little task force to help organize these briefings, and have your input on how much time  
3 to devote to it and so forth.

4 DR. McCABE: Does anyone wish to volunteer? Barbara, Debra, Agnes,  
5 Emily. Yes, please. We'll call on the ad hocs probably as well, but thank you to the two of you  
6 for volunteering, and Paul. You have been a leader in this in the EEOC. We may call on others  
7 of you as needed, especially among the ad hocs. If anybody has been shy and wishes to  
8 volunteer, at the break, please let Sarah know.

9 Dr. Boughman, thank you very much for briefing us, and thank you for your  
10 efforts. They clearly fit with those expressed by this committee in our correspondence in the  
11 past.

12 So next, we're actually running just a little bit ahead of time, but we're going  
13 to have a presentation on information gathered on efforts in genetics education and training by  
14 Dr. Joan Reede. Dr. Reede will review the education task force's information gathering efforts  
15 over the last three months.

16 I'd like to thank Joan for your chairing the task force, as well as Kim  
17 Zellmer, Hunt Willard, Barbara Harrison, and Agnes Masny for your service on the task force.  
18 We appreciate the time and effort that all of you have put into gathering the data and preparing  
19 the draft resolution.

20 Joan, please proceed.

21 DR. REEDE: Thank you very much, and thank you for the opportunity to  
22 present the work of the task force.

23 You just mentioned the names of the members of that task force, but I also  
24 want to acknowledge the staff who supported us. Amanda Sarata did a wonderful job of  
25 helping to facilitate our meetings, moving this forward, and helping to put this presentation  
26 together.

27 During the discussions of genetic education and training at the March  
28 meeting, the committee decided to draft a resolution to the Secretary on the issue of genetics  
29 education and training, outlining key recommendations in this area. Toward this end, they  
30 decided that a task force should be established with these charges to collect information on the  
31 activities of health professional organizations that relate to genetics education and training, to  
32 organize and facilitate a roundtable discussion to be held during this June meeting, and to draft  
33 a resolution to the Secretary on genetics education and training.

34 The committee's request to hear formally from professional organizations in  
35 the private sector on their activities in genetics education and training was meant to serve as a  
36 follow up to, or addition to the survey of federal agencies on their activities, as well as the  
37 presentation and information from Joann Boughman at the October meeting.

38 It was felt that this information would be used to inform our resolution, and  
39 should ensure that any recommendations made to the federal government would be  
40 complementary to activities already ongoing in the private sector. We solicited information  
41 from 26 organizations.

42 The organizations can be divided into three categories. Genetic-specific  
43 organizations, health professional educational organizations, organizations involved in the  
44 education of health professionals, and health profession organizations. The latter really  
45 targeting three groups, those that represent generalists such as AMA, those representing

1 specialities such as family practitioners, pediatricians, OB/GYN, and those representing  
2 specific constituencies, such as the National Medical Association, AAIP, and NHMA.

3 Of note, we had only a maximum of nine organizations in each category,  
4 and this was really to be consistent with guidelines so that we would not have to turn to OMB  
5 for review of our survey of the organizations. Fifteen of the 26 organizations responded, which  
6 is a fairly good response for a three-week period from when the surveys went out. Of note,  
7 however, we shouldn't assume that the groups that we did not hear from do not have important  
8 activities in these areas.

9 Here is a list of the organizations that responded. You'll be hearing from  
10 some of these organizations during the roundtable discussion, as well as some additional  
11 organizations that did not get their responses in in time for this.

12 In addition to having information that represents organizations, it is also  
13 important to note that this information represents multiple specialties or disciplines. There is  
14 information on nurses, geneticists, allied health professionals, dentists, pharmacists, and  
15 physicians.

16 With regard to the three categories, I will go through them in sequence. The  
17 first is on genetic-specific organizations. The committee felt that it was important to solicit  
18 information from organizations who are focused on the issues of genetics. This provided the  
19 committee with the information about the "state of the state," which is currently going on  
20 within organizations that have a high awareness of and focus on genetics and genomics.

21 We have already benefitted greatly from their input on many issues, and  
22 appreciate their willingness to continue providing their input. We highlighted three important  
23 areas of potential gaps in our understanding. These were diversity, life insurance certification,  
24 and curricular development.

25 We wanted to know about current initiatives and activities that related to  
26 diversity in the genetics workforce, that enhanced genetics and genomics curricula, and that  
27 promoted the incorporation of genetics and genomics content into licensure and certification.

28 Diversity in the genetics work force is an important issue to the committee.  
29 Genetics is a field where both cultural diversity and cultural competency are particularly  
30 relevant, the impact of culture on perceptions of attitudes about genetics, and also in the area of  
31 the debate about the scientific basis of race and its place in medicine and genetics.

32 What we found was that these organizations were involved in many  
33 activities that targeted both the pipeline, such as those involving K through 12 education, as  
34 well as the recruitment of individuals from diverse backgrounds into genetic counseling.

35 These efforts also targeted minority health professionals, minorities who  
36 attended colleges, high schools, colleges and universities, and also at the organizational level  
37 brought diversity in through structural forms, such as the organization of a diversity standing  
38 subcommittee.

39 With regard to curricula, it appears that genetic-specific groups are leaders  
40 in the development of genetic curriculum, and are actively engaged in dissemination and  
41 outreach efforts to other health professionals. They are engaged in activities such as speakers  
42 bureaus, short courses, conferences, as well as the dissemination of position papers and practice  
43 guidelines.

44 The core competencies provide universal guidance, and the core  
45 competencies have been a major effort on the part of these genetic-specific organizations. It

1 was felt that specialty-specific competencies could be determined by professional societies, and  
2 the use of the Internet to disseminate and share information was also felt to be critical.

3 Increasingly, medicine will become more electronic, and genetics, being a  
4 field that changes rapidly and involves large amounts of information, benefits from this change.  
5 It was felt that educational materials should conform to this trend.

6 Courses to train faculty to teach genetics content is a key element. This  
7 matches up with a barrier that was noted by another category, the Health Professional  
8 Educational Organizations, feeling that there was a lack of appropriately prepared faculty for  
9 genetics training and education.

10 Licensure, certification, and accreditation have the potential to change  
11 behavior, and increase integration of genetics and genomics knowledge throughout the health  
12 care system. Although many of the agencies did not report a great deal of activity in these  
13 areas, some of the types of activities that they are engaged in included credentialing programs  
14 in genetics, use of core competencies to help inform discussions or programs related to  
15 licensure and certification, and biannual genetics review courses.

16 The issue of licensure certification and accreditation can require several  
17 different types of specific policy solutions. Part of this is related to the fact that there are  
18 different factors across the board here. In licensure, oftentimes we're dealing with state  
19 certifications, we're dealing with specialty professional societies in the federal government such  
20 as with CLIA, and accreditation, and we're dealing with private organizations, such as JCAHO.

21 The overall recommendation from this group was that "we should be strong  
22 in our appeal to helping the HHS Secretary to actively support a wide variety of endeavors  
23 based in or funded by any of the HHS-based agencies, as well as seeking partnerships with  
24 other relevant federal agencies."

25 The second category. Those organizations that are involved in the  
26 education of health professionals. For this category, these questions deal with curricula  
27 development for education of health professionals. With these questions, we are trying to  
28 gauge where genetics/genomics stands in health education organizations that have a general,  
29 rather than a specific, genetics focus.

30 The question for this group included those about the need for the integration  
31 of genetics and genomics in their curriculum, barriers to this integration, and current ongoing  
32 activities and initiatives.

33 With regard to their perceived needs, there is a need to acknowledge that  
34 genetic science is for both generalists and specialists. This points out the tension between  
35 educating everyone, and maintaining a niche for specialists. General practitioners have an  
36 important role to play in integrating genetics and genomics in health care, and specialists will  
37 also play a necessary role in that new paradigm.

38 Due to the complexity of and the speed at which the genetics field is  
39 evolving, there is also a need for improved access to knowledge, and this was cited by many  
40 organizations. There is a need to be able to evaluate product claims. This comment ties to the  
41 committee's interest in and focus on direct-to-consumer marketing and advertising. Education  
42 will facilitate provider's ability to assess the validity, efficacy, and safety of various new  
43 products for their patients.

44 There is a need to redefine and recast genetics as an inherent and  
45 overarching part of health and to improve communication between all health professionals and

1 the public about how genetics affects health. In keeping with the theme that genetics is relevant  
2 to all specialties in medicine, and will be an important part of all aspects of clinical  
3 management of the patient, from prevention, diagnosis, and treatment perspectives.

4 There was a perceived need to determine the level of knowledge that is  
5 needed, who needs to know what, and who should provide that, and also the need to provide  
6 tools for lifelong learning. This last comment highlights the fact that genetics and genomics  
7 knowledge will be rapidly changing, and therefore, education should be a lifelong pursuit.

8 With regard to barriers, it was difficult to find current case examples or  
9 models. This comment represents an important theme that was raised by several organizations  
10 in response to many different questions. In order to teach genetics and genomics, models using  
11 clinically relevant examples are needed. There is a lack of trained faculty broadly competent in  
12 genetics and genomics, and it is expected that there will be a lack of uniformity in the rate of  
13 integration of genetics into the various specialties in medicine. This, in turn, will make it  
14 difficult to determine who should be learning what, and when.

15 Other barriers include difficulty in motivating students to learn something  
16 based on the promise of its "future importance," an overcrowded curricula that is already  
17 struggling with issues of basic science and clinical practice, and the fact that genetics by many  
18 is still considered to be an esoteric field.

19 Current activities from health professional educational organizations  
20 include membership in NCHPEG, integrating those core competencies that have been identified  
21 by NCHPEG into their entry-level competencies for their specialties, or their special  
22 disciplines, and the serving of faculty competency in and understanding of genetics/genomics.

23 With regard to current activities, one organization is sponsoring relevant  
24 legislation of the allied health professions that would help support curricular development in  
25 the area of genetics and genomics. Overall, it was felt that cultural competency was addressed  
26 broadly within the context of outreach to underserved populations, and there was no specific  
27 focus on the area of genetics.

28 Recommendations from these organizations included that schools and  
29 professional organizations must provide leadership in preparing the next generation of health  
30 professionals in genetics. The primary role of preparing health professionals lies with the  
31 schools and professional organizations, not with the federal government.

32 Continuing education is needed to train the trainer, and this specifically  
33 refers to helping clinicians to determine when to refer, how to obtain information about  
34 genetics research studies, and ways to discuss research options with patients. More funding is  
35 needed to support training and education in genetic technologies, and to facilitate the  
36 incorporation of new knowledge and skills.

37 With regard to the last set of organizations, the health professional  
38 organization responses, the committee reasoned that it was also important to gather information  
39 from umbrella organizations that focused on specific health professional disciplines. These  
40 responses provide the committee with useful information on the relative importance of genetics  
41 to organizations grappling with many other equally important issues.

42 The information may also be an indicator of how genetics and genomics is  
43 perceived within the health care system generally. The types of questions that were asked of  
44 this group include characterizing the need for integration of genetics and genomics, what types  
45 of activities or initiatives they have currently ongoing, and in here, we ask specifically about

1 partnerships, about interdisciplinary efforts, about educational products, and about outcomes,  
2 what has been evaluated, and the impact.

3 We asked them specifically about steps they were taking with regard to  
4 diversity, and about their particular concerns and recommendations that they would like our  
5 committee to make. With regards to their needs, they felt that all health professionals need a  
6 strong, knowledge-base in genetics and genetics testing.

7 Identifying clinically-relevant examples of genetics and genomics would  
8 help address the need to understand how genetics interfaces with practice. You hear a  
9 recurring theme of the need for applications, and there is a need to understand how this relates  
10 to practice.

11 There is a need to help professionals with  
12 up-to-date advances in genetics. This is brought by many health professional organizations in  
13 terms of the need for access to new knowledge. Again, we have the issue of lifelong learning,  
14 keeping up to date. In terms of current activities, the health professional organizations are  
15 undertaking a broad array of activities related to genetics education and training. Some are  
16 using traditional tools for consensus building and dissemination. You see a wide range from  
17 CME educational sessions, to web-based educational tools, to newspaper articles, journal  
18 articles, symposia, and across the board.

19 With regards to their current activities and their interdisciplinary nature or  
20 partnerships, several of the organizations are partnering with others, federal agencies, genetic  
21 specialty societies, medical societies, non-profit organizations, and private companies. The  
22 majority of the organizations report that their activities are interdisciplinary in nature.

23 With regards to outcome and evaluation, it was found that most of the  
24 organizations measured their outcomes or evaluation based on increasing interest, or continued  
25 interest, and in issues related to genetics and genomics. This was gathered by looking at  
26 numbers who attended genetic sessions at national meetings, hits to genetics websites, CME  
27 certificates, and distribution of educational materials.

28 Current activities related to diversity take many forms, from community  
29 outreach, career development, education, research, advocacy, and organizational position  
30 statements. Concerns of this last group of organizations. The science underlying issues of race  
31 in medicine needs to be examined and integrated into genetics education and training.

32 Issues of race in genetics necessitate special consideration and treatment in  
33 the educational setting. Awareness of these issues is an important part of a health practitioner's  
34 cultural competency. For many physicians, genetics does not have immediate, daily, and  
35 clinical applicability.

36 This comment relates to the need to understand how genetics interfaces with  
37 practice, and the barriers caused by the difficulty in finding case examples or models in  
38 genetics and genomics cited by health professional organizations previously.

39 To quote one person, "Genomics is spinach. Everybody knows it is good  
40 for them, but nobody likes it." Genetics education must be represented throughout the entire  
41 continuum of medical education. There is a need for tools that facilitate this lifelong learning,  
42 and a need to keep professionals up to date.

43 Additional concerns. Educational programs must have a focus on pediatrics  
44 and genetics, and physicians prefer interactive learning with case studies. The effect of the  
45 nursing shortage on patient education and informed consent was also raised. It was felt that

1 this point might raise the broader question of the impact with the emphasis on efficacy in our  
2 system might have on integration of a rather time intensive and complex issue, like genomics.

3           Recommendations from this third set of organizations. Ongoing, continuing  
4 education should be the responsibility of the licensing agencies and professional organizations.  
5 The development of profession-specific materials should be left to the health professions. The  
6 support for genetic education programs would be an appropriate role for the government.

7           Further recommendations. Funding is needed for new programs that feature  
8 educational practices known to change physician behavior. This includes areas such as  
9 learning based on perceived need, and interactive learning. Efforts in genetics education must  
10 extend to related areas of molecular medicine, and benchmarks should be instituted to inform  
11 and assess the bidirectional impact of translational research, i.e. from bench to bedside, and  
12 bedside to bench.

13           Recommendations. Education and training should address population-based  
14 genetic variation, and its utility in the emerging era of individualized medicine. The impact of  
15 genetic polymorphisms on the determination of "what is normal" in the era of molecular  
16 medicine, the scientific relevance and significance of minority participation in clinical trials, to  
17 the quality of health care in a merging era of genomic medicine, and the diagnostic importance  
18 of obtaining a good and complete family history on all patients.

19           Turning now to our second charge, which relates to the educational  
20 roundtable. The purpose and goals of this were to discuss in detail, the organization's efforts in  
21 and attitudes about genetics education and training, and to add to the information already  
22 provided in the surveys, to identify key concerns and barriers with respect to these issues on the  
23 organizational level, and to help inform any resolution coming from this committee to the  
24 Secretary.

25 The organizations that will be involved in this roundtable are listed here. Our representatives  
26 will be introduced to you formally later.

27           In the third area, we were asked to develop a draft resolution that could go  
28 to the Secretary with regards to education and training. The purpose is to arrive at a consensus  
29 of the committee on the issue of education and training, to convey this consensus to the  
30 Secretary, and to make recommendations or possible steps to address this important issue.

31           The conclusions that we had were that genomics can improve health, that  
32 adequate education and training in genetics and genomics is essential to integrating genetics  
33 into the health care system, and that access is contingent upon effective integration.

34           Additionally, the education professional organizations identified the  
35 following needs. There is a need for inventoried, widely-relevant clinical applications, a  
36 recurring theme, educational models that use such applications, and a broadened focus from  
37 genetics to genomics. There is also a need for appropriately trained faculty, and training  
38 programs that address genetics/genomics, and public policy.

39           With regard to our task force education resolution recommendations, there  
40 were six. We decided that while genetics is important and special, in that it is relevant to all  
41 areas of medicine and health care, it would be inappropriate to single it out in the educational  
42 setting. Rather, it should be integrated throughout all stages of learning, in all settings, and  
43 throughout all disciplines.

44           There is a need to support programs that enhance diversity among cultural  
45 competency of health professions. We need to engage other stakeholders in the process of

1 cataloging genomics applications to clinical medicine and public health. This third  
2 recommendation addresses the concern about the lack of clinically relevant genomics and  
3 genetics applications.

4 Fourth, we need to support programs that "train the trainers" in genomics  
5 and genetics education, addressing the identification of barriers to the lack of professionals  
6 trained to teach genetics. Fifth, we need to promote communication between faculty to  
7 enhance use of genomics educational models. And last, to encourage incorporation of genetics  
8 and genomics into the certification and licensure process.

9 Thank you.

10 DR. McCABE: Thank you very much, Dr. Reede.

11 At this time, let's take a 15-minute break. The members of the committee  
12 and our ex officios are invited to enjoy the refreshments here at the front of the room. For  
13 members of the public, refreshments and beverages are available at the gift shop near the hotel  
14 lobby.

15 We will reconvene in 15 minutes. That will be at about five of the hour.

16 Thank you.

17 (Recess.)

18 DR. McCABE: If everybody could take your seats, please. We're now  
19 going to proceed with a roundtable discussion with the eight organizations on their efforts in  
20 educating and training health professionals in genetics.

21 The purpose of this roundtable discussion is to explore in greater depth the  
22 efforts of key professional societies and educational organizations to enhance knowledge of  
23 health professionals in genetics and genetic technologies, and to identify what steps, if any,  
24 need to be taken to advance these efforts.

25 At this time, I'd like to welcome our roundtable of participants. You've  
26 already joined us at the table, so I don't need to extend that invitation.

27 I'll now turn over the gavel to Dr. Reede, who will lead the discussion and  
28 introduce the roundtable members, or have them introduce themselves.

29 Joan?

30 DR. REEDE: Thank you, and thank you very much to those who are  
31 participating in this roundtable discussion.

32 I'm going to introduce them, and they're not in order. They're Joann  
33 Boughman, who is the Executive Vice President of the American Society of Human Genetics, a  
34 medical geneticist and adjunct professor in the Departments of Pediatrics, Obstetrics,  
35 Gynecology, and Reproductive Sciences at the University of Maryland.

36 Joe McInerney, who is the Director of the National Coalition for Health  
37 Professional Education in Genetics. He was also, in the past, Director of the Foundation for  
38 Genetics Education and Counseling.

39 Felissa Lashley, and Dr. Lashley is Dean of the College of Nursing at  
40 Rutgers, the State University of New Jersey. Previously, she was Dean and Professor at  
41 Southern Illinois University School of Nursing, and a clinical Professor of Pediatrics at the  
42 University's School of Medicine.

43 Dawn Allain, who is a genetic counselor for the Children's Hospital of  
44 Wisconsin in Milwaukee. She is a genetic counselor and clinic coordinator for the Cancer  
45 Genetic Screening Program at Froedtert Hospital in Milwaukee, and is the current President of

1 the National Society of Genetic Counselors.

2 Judith Lewis. Dr. Lewis is a professor in the Maternal Child Nursing  
3 Department at Virginia Commonwealth University, and also Director of Information  
4 Technology for the School of Nursing.

5 Georgia Dunston. Dr. Dunston is professor and chair of the Department of  
6 Microbiology at Howard University College of Medicine and founding director of the newly  
7 formed National Human Genome Center at Howard University. She is here representing the  
8 National Medical Association.

9 Toby Citrin, who is Director of the Office of Community-Based Public  
10 Health at the University of Michigan School of Public Health and Director of the Michigan  
11 Center for Genomics and Public Health. He is here representing the Association of Schools of  
12 Public Health.

13 Michael Whitcomb. Dr. Whitcomb is Senior Vice President of the  
14 Association of American Medical Colleges, and in the past served two terms as Dean at one,  
15 the University of Missouri Columbia, and the other at the University of Washington.

16 Thank you all for being here.

17 I'd like to open it up with a question for all of you. If you could speak a  
18 little bit to your key issues, concerns, and problems with respect to genetics education and  
19 training from the perspective of your organizations. Here, if you could in particular refer to  
20 gaps in current genetics education of health professionals.  
21 Would one of you like to take it? Mr. Citrin?

22 MR. CITRIN: I'll take a stab at it. I should say initially at the outset that to  
23 my knowledge, the Association for Schools of Public Health, who I'm here today representing,  
24 has not a current survey of what the schools in fact are doing in incorporating genomics into  
25 education. But we did have a graduate student of ours do a survey based on websites of all 35  
26 schools of Public Health to see the extent to which genomics has found its way into programs  
27 or curricula. I shared that survey with Amanda Sarata, for whatever usefulness it might have to  
28 your work.

29 Interestingly enough, of the 35 schools, ten of the schools have been stating  
30 that they now have genetics programs, so most of these are very research-related, as opposed to  
31 teaching related. Twelve schools are offering courses. Very few schools are identifying  
32 ethical/legal/social issues among what they are teaching. So the current scene, if that is an  
33 indication, suggests that the large majority of schools of public health really have not  
34 incorporated genetics or genomics into the content of the education of people going into public  
35 health.

36 This, in spite of the fact, or maybe leading to the fact that the Institute of  
37 Medicine's report on the teaching of public health that came out in 2003, "Who Will Keep the  
38 Public Healthy?" makes a strong plea for genomics being one of the eight content areas that all  
39 schools of public health should teach.

40 Now, with specific reference to your question of one of the barriers. The  
41 very fact that the recommendation of the IOM report is that these eight content areas should  
42 now be incorporated into the teaching of public health, rather than replacing what is now being  
43 taught, suggests an add-on to what is already seen by many in the schools as an overload of  
44 information to be taught to students.

45 Now, the answer to that question, of course, is integration. In fact, the



1 "Who Will Keep the Public Healthy?" report suggests that genomics needs to be integrated into  
2 an overall ecological view of causation of health and disease. But this suggests that people  
3 who teach epidemiology, biostatistics, health policy, and environmental health and health  
4 behavior, should all incorporate genomics in what they teach. Most of the people who teach  
5 these subjects do not understand the role that genomics plays in these areas, and the field has  
6 been so fast moving, that it is hard to have people who specialize in these, what are now  
7 identified areas of public health, to keep in touch.

8 So most of the schools that say they are teaching genetics, these 10 or 12,  
9 have specific, discreet courses that are typically electives, so that by far, students going through  
10 schools of public health today are receiving little or no genetics education. So the ability to  
11 have people who know how to teach incorporating genomics into the broader view of causation  
12 of health and disease, this would be a very major barrier.

13 The issue of time to incorporate this into what is already a very busy  
14 curriculum is yet another. Of course, related to that is the fact that there is no requirement to  
15 incorporate genetics, and it was good to see that accrediting bodies are incorporated into the  
16 draft resolution that you have put together.

17 So these, it seems to me, are some of the barriers. I want to add another  
18 barrier with respect to the one aspect of teaching, which is the ethical/legal/social issues. The  
19 very teaching of ethics in schools of public health is a hot topic these days. Most schools do  
20 not require training in ethics as a condition of getting a masters degree in public health. So if  
21 ethical/legal/social issues related to genetics are to be part of the required curriculum, which  
22 many of us who are in this field feel it should be, this then presupposes that ethics itself is a  
23 part of the curriculum of people going through schools of public health, it since becomes part  
24 of a broader issue. So that is at least one summary of some of the issues that colleagues of  
25 ours, and people in our school are seeing.

26 DR. REEDE: Thank you very much.

27 Dr. Lashley?

28 DR. LASHLEY: I'm here representing AACN, which is the national voice  
29 for baccalaureate and higher degree in nursing programs across the country. The membership  
30 consists of the deans and directors of those programs throughout the country.

31 AACN has had a few efforts in recognizing genetics as an essential part of  
32 nursing curricula. One of the first was to incorporate some knowledge of genetics in the 1998  
33 "Essentials of Baccalaureate Education" document that was put out by the group. What this  
34 group does is put out essentials for the different levels of education that includes core content  
35 and core competencies for nursing at the baccalaureate and at the higher degree levels.

36 At the masters level, there was no such content included when that  
37 document came out originally, but that is going to be revised, and provides an opportunity for  
38 getting those essentials into those curricula as well, and that would be advanced practice  
39 nursing and nurse practitioner programs, and the like.

40 They have also had some conferences on incorporating genetics, one in  
41 which Dr. Collins spoke to the group, and I did as well, as far as applying genetics education  
42 into nursing curricula. So the group is moving on the need for that. What some of the barriers  
43 are are similar, I think, to what all the health professions are experiencing, and those are the  
44 intensity and density of the curriculum. There are a lot of competing topics for all of us in  
45 terms of what we put into our programs, and how much time we spend on them.

1                   The second is that there has been traditionally a lot that relates to genetics  
2 in nursing programs throughout the years. But they tend to be more in the specialized, discreet  
3 disease-type areas, rather than a broad look at the influence of genetic and genomics across the  
4 curriculum, where most of us believe that that needs to be in terms of patient education,  
5 counseling, and that those things are not all that different for people with genetic disorders, but  
6 that the knowledge has to be integrated throughout the nursing curriculum.

7                   Another barrier has been the lack of qualified faculty to be able to do that,  
8 and there have been efforts to address that. Summer institutes, NINR programs, in terms of  
9 building that out, but it hasn't suffused the programs, if you will, nationally. So I think those  
10 are some of our major challenges, is getting genetics through the life span in nursing education.

11                   DR. REEDE: Thank you.

12                   Dr. Lewis?

13                   DR. LEWIS: Probably a nice segue from the same discipline, and I'm here  
14 today as President of the International Society of Nurses in Genetics. It is also interesting to  
15 me, having been a member of the former Secretary's Advisory Committee on Genetic Testing,  
16 to watch the continued growth and development, and I want to applaud you all for continuing  
17 along some of the lines that we felt were so important.

18                   One of the things that I really think is important is when you talked about  
19 the lack of looking at genetics as exceptional. I think that becomes really important. If it  
20 becomes part of base knowledge, rather than considered specialty knowledge, that is very  
21 critical. I think of myself when I worked as an advanced practice nurse, and if every time I saw  
22 a patient with an elevated temperature, I called in the infectious disease specialist, we wouldn't  
23 get very far.

24                   The infectious disease specialist certainly has a place in looking at patients  
25 who have complicated problems, but when I think we get to the point where every generalist is  
26 able to incorporate basic genetic knowledge, and those of us with specialty knowledge become  
27 like the infectious disease specialist, or the subspecialist, that that is when we'll know we've  
28 arrived. So part of what ISONG is working really hard to do is to ensure that all clinicians and  
29 all health care professionals, and in our case, we can only deal with nurses, but that people have  
30 a certain base knowledge.

31                   ISONG has published in collaboration with the American Nurse's  
32 Association, scope and standards of practice of looking at what kind of knowledge is based on  
33 the NCHPEG competencies. But looking at what kind of knowledge we would expect a  
34 generalist to have, and what kind of knowledge we would expect a specialist to have, that  
35 model makes an awful lot of sense. And because we are a relatively small group of people who  
36 are specialists, part of our goal for ISONG is to work with our 2.7 million colleagues who are  
37 not members of ISONG, to ensure that people have basic knowledge, and that we're available as  
38 consultants and collaborators, but power is only powerful when you share it. So the idea of  
39 being able to share it is so that the knowledge base trickles down.

40                   What are the barriers to that? The barriers are, I think the same ones that  
41 have been recognized in terms of competing priorities for all of us. But I also am old enough to  
42 remember when the new disease called HIV was discovered in the early '80s, and we got  
43 mandates to include that in the curriculum, and we sat there and argued over what was going to  
44 stay in and what was going to go out, so we could include information on HIV and AIDS.  
45 Somehow we have managed to do that, and I don't know of a health professions education that

1 doesn't include content on HIV at this point.

2           So I think it can be done, it is just a matter of making sure that everybody  
3 has the will. So I'm really pleased to see the certification and the credentialing bodies at the  
4 table, because I think that is where the push is going to come. Good will only goes so far, and  
5 mandates work much better.

6           DR. REEDE: Thank you.

7           Dr. Whitcomb?

8           DR. WHITCOMB: Well, from the perspective of medicine, it may surprise  
9 some of you when I say that I really don't think that the issue, in terms of being a barrier, really  
10 relates to how crowded the curriculum is. I think that the issues, as they relate to medicine, are  
11 two fundamental problems that sort of transcend all education, at least at the level of thinking  
12 about the continuum of education, including medical school, and then onto residency training.

13           One of those is simply how do you integrate content that should be  
14 integrated really across the curriculum in a way that makes that content relevant, important, and  
15 learnable? We are perhaps fortunate in one sense, because the movement in medical education  
16 reform, as it relates to the undergraduate, medical school curriculum for the past decade has  
17 really been to evolve towards an integrated curriculum, as opposed to the  
18 discipline-specific sort of departmentally-controlled courses that were typical of most medical  
19 school curricula.

20           This is an ongoing process with many schools involved, and it takes time to  
21 do it, but I think the challenge is simply then how do you integrate within the framework of that  
22 curriculum? I think perhaps the biggest challenge as we look down the road, and I think this  
23 will apply not only to medical students, but I think it will be an issue for residency education as  
24 well, is the opportunity for the learners to have exposure to patients that will make real for them  
25 what it is they are expected to learn, and how they will then apply their learning in the patient  
26 care environment.

27           That is an extraordinary challenge of an increasing severity simply for  
28 education of medical students at all levels. I think that with the changes that are occurring  
29 within the delivery system, it will continue to be a major challenge as we try to figure out how  
30 to get access to patients, the kinds of patients that students and residents need to be exposed to,  
31 since more and more of that care is being conducted outside of the traditional teaching site,  
32 which is the major teaching hospitals. So I really think those are the two major barriers right  
33 now for medicine.

34           DR. REEDE: Thank you.

35           Mr. McInerney?

36           MR. McINERNEY: Thank you. The barriers that everyone has discussed  
37 so far, apply across the board for NCHPEG's efforts. We have more than 145 member  
38 organizations now here in the United States, and abroad as well. Certainly we have a fair  
39 number of organizations that represent physicians, but the vast majority of our members are  
40 non-physician allied health groups, commercial organizations, consumer groups, and so on.

41           So one of our great challenges is to address the needs of this extraordinarily  
42 diverse membership. I should say that across the board, again, the greatest gap we face is the  
43 availability of evidence and educational materials that demonstrate to our constituents that  
44 genetics makes a difference in patient outcome, and can change effectively what they do now,  
45 rather than five years from now. We need to make that case more dramatically, we need the

1 evidence to do that, and we need the educational materials that can do that.

2 Another gap is providing more guidance about what to teach. You have  
3 heard referenced a number of times this morning for the core competencies, and I think they  
4 have been very effective. Lots of organizations have begun to incorporate the core  
5 competencies into the development of their own curricula, whether it is undergraduate  
6 curriculum, or continuing education.

7 Increasingly, we are getting questions about what should we teach then?  
8 What is the content that we should teach? I think the core competencies do a very nice job of  
9 saying this is where we would like your constituents to end up when they're finished, but what  
10 do we teach to help get them there?

11 This week, we're posting on the NCHPEG website a set of core principles in  
12 genetics that really are, we believe, core concepts, that most health professionals should be able  
13 to understand with respect to basic genetics. So that's another gap.

14 Another serious gap for us, and we're trying, believe me, we're trying, is the  
15 issue of diversity and cultural competence. How do we represent issues related to diversity and  
16 cultural competence more effectively in our programs, and how do we involve organizations  
17 more effectively that can help us do that? So any suggestions that anybody in this room has to  
18 help us do that more effectively, we would certainly appreciate.

19 The last gap for us, I think, is related to what I have just mentioned. That is,  
20 there may be some significant gaps in our membership. There may be entire constituencies  
21 within the health professions that we are not reaching, that we need to be reaching, and that we  
22 need to have involved in the development of our materials.

23 We have a membership committee that is looking at that issue now, taking a  
24 look at our membership and how it sorts out with respect to disciplines, and trying to figure out  
25 where we should place our efforts in that regard.

26 DR. REEDE: Thank you very much.

27 One of the recurring themes for many of you related to faculty, and the lack  
28 of trained faculty in genetics as a barrier. Do you have suggestions on how we could address  
29 this issue of training our faculty? Dr. Lewis?

30 DR. LEWIS: There are several programs that I know of that are available  
31 that might serve as models in terms of nursing. I believe one of the ELSI-funded projects was  
32 the project that Cindy Prows has out of Cincinnati Children's Medical Center that runs in two  
33 formats. One is web-based for 16 or 18 weeks, and the other is a summer, two-week intensive  
34 program that is basically designed to provide nursing faculty who have no previous knowledge,  
35 or have minimal knowledge in genetics, with what they need to infuse the curricula with  
36 genetics.

37 I took that a couple of years ago, and every year I get a follow-up server that  
38 said so, what did you do this year? You took this course and you used the government's money,  
39 and what have you done this year? That is one program that I know of that exists that was  
40 ELSI-funded, I believe. I may be wrong in that, but I'm pretty sure that is where the original  
41 funding came from.

42 Dale Lea at the Foundation for Blood Research has put out a group of  
43 modules that are available that you can order by mail that give faculty information that they can  
44 then use and adapt to their curricula. Then there are other programs like the National Institute  
45 of Nursing Research has the summer genetics institute that is an

1 8-week residential program for advanced practice nurses, and nurse faculty that deal with both  
2 education and research training. That one is really to help faculty with the knowledge  
3 development, as well as the knowledge transmission role.

4 I think there is some really good programs out there. The problem is that at  
5 least in nursing, they are relatively small. My class last summer at SGI was 20, and we were  
6 the fourth or fifth class. So in five years, they have trained 100 nurse educators. The  
7 Cincinnati program probably has groups of 25 twice a year, so I think there are programs there.  
8 Part of it is volume, part of it is taking people and finding people who are willing to do this,  
9 and then get back to their home institutions, and then have to convince curriculum committees  
10 that this material becomes important.

11 So I think there are models out there, but the question is what is the best  
12 model? I think a mixed model is best. Some people learn best in web-based, other people learn  
13 best when you take them out of their environment for a couple of weeks and turn off their  
14 pagers and their cell phones. So I think it is just a matter of what works best for individuals,  
15 but there are models out there, at least that I know of in my discipline.

16 DR. REEDE: Mr. McInerney?

17 MR. McINERNEY: Yes, thank you. I wanted to comment on one of the  
18 slides you showed during your presentation, and the comment that physicians prefer interactive  
19 learning with case studies. I can tell you from our experience with the NCHPEG membership  
20 that that applies across the board to all health professionals, not just physicians. We found that  
21 that is probably the best way to go, to engage people in the genetics content.

22 With respect to models, we have just finished developing in conjunction  
23 with a number of colleagues from the dental community, a program to train faculty in dentistry  
24 and dental hygienics. It is creatively titled "Genetics, Dentistry, and Health" on our website.  
25 Go to [nchpeg.org](http://nchpeg.org) and you can find that.

26 We have developed that around case studies with the intent of trying to get  
27 dentists and dental hygienists ultimately to change practice, to think a little bit differently about  
28 genetics when their patients come to them. But again, it is focused on case studies, with some  
29 core basic genetics.

30 DR. REEDE: Dr. Dunston?

31 DR. DUNSTON: First, just to set the record straight, I am a former chair of  
32 the Department of Microbiology as of March of this year. Also, I'm sitting in for Dr. Randall  
33 Maxey, the President of the National Medical Association. I just wanted to make a comment to  
34 your first point, and then the comment on education.

35 The National Medical Association promotes the collective interests of  
36 physicians and patients of African descent. It is the largest and oldest national organization  
37 representing the interests of African Americans, representing more than 25,000 physicians, and  
38 the patients that they serve.

39 The NMA is dedicated to keeping its members abreast of the many rapidly  
40 occurring advances across the various medical specialties, and the other changes affecting  
41 medical practice. So from that general perspective, certainly the organization considers  
42 genetics and genomics of paramount importance.

43 The National Medical Association was founded more than 100 years ago,  
44 and had its formation in the history of the American community. From that history, the subject  
45 of genetics becomes particularly important, because the subject concerns the inclusion of

1 African Americans, not just as a social imperative, but the subject matter forces the inclusion of  
2 African Americans as a scientific imperative, recognizing that the subject of genetics itself is  
3 challenging to our whole concept, construct, as well as methods of teaching.

4 Biology. We are moving from group-based kind of analyses, to  
5 individualized. Genetics as a discipline is often equated with the prospects of individualizing  
6 medicine. So there is a particular challenge that the NMA recognizes in a science that has the  
7 potential of individualizing medicine in a society that tends to group in its approach and  
8 methodology. So genetics as a subject that deals with biological identification, and  
9 classification of groups at all levels, is particularly challenging.

10 The issue, as was stated on one of your slides, is what does the science of  
11 genetics have to say about our biological thinking about race, race medicine, profiling in  
12 medicine, and developing drugs for groups when you are using a science that is potentially  
13 distinct in its capacity to individualize?

14 The NMA is sensitive to genetics as a subject that is very destabilizing in its  
15 whole concept with regard to the community. In the evolution of the science, the organization  
16 is concerned about the apprehensions of the community to become engaged in the potential  
17 benefits because of the historical context, in which the community, as well as the particular  
18 science, has been applied.

19 So a major barrier is how do we engage the community in such a way that  
20 the prospects to benefit become the driving force? To be proactive for engagement,  
21 recognizing that exclusion is not an option in the science of genetics? Because the science  
22 itself deals with the fundamentals of disease diagnosis, treatment, and progression in ways that  
23 not being included at every level of the science and its application in society is critical to how  
24 help will be administered to the group as long as we are part of this American community.

25 So the education of the community, as well as the professionals in medicine  
26 is of paramount importance, as reflected in the meetings at the local, regional, and national  
27 level that the organization sponsors in its continuing medical education, and its outreach to  
28 community. It is outreach to the community in ways that are complemented by current focus on  
29 faith-based organizations. Again, the history of the population is one where the engagement of  
30 faith-based organizations become perhaps a proactive means of addressing a subject that tends  
31 to be approached from a fear-based perspective.

32 So the challenge of how do you ensure that as new knowledge is coming  
33 forth, that you are represented, that your perspective is reflected, is paramount, and is certainly  
34 underscored. I would just say as one who is in academia, the role of education about the  
35 science, as well as the application of the science, and helping the community to appreciate that,  
36 that this is a science that demands participation.

37 It is a science that is challenging the reference point, and is challenging  
38 definition of what is norm. Inclusion of African people brings with it the challenge of variation  
39 and diversity in biology, and needs to recognize that what one sees cannot be separated from  
40 the context and the reference point from which you are viewing. How do we begin to  
41 incorporate that in our medical practice? That we have diversity as a strength, as an instructive  
42 commodity, not just a pathological or a basis of disease. That is my last point.

43 In trying to engage the community in terms of genetics and its impact,  
44 recognizing that the genome project not only led us to techniques and technologies for  
45 identifying single genes that have major effects in causing disease, but it now has brought us to

1 an appreciation of haplotype variation, and many common genes that vary, but are not  
2 pathological, just different in their functional capacities, and that they work together to  
3 accomplish functions.

4 So how do we begin to change the concept of medicine when the gene is not  
5 the pathological agent, but it is a means by which we can analyze, process, and understand  
6 disease, not always as a structural defect, but a regulatory one, and understand what is  
7 regulating the expression of genes that are associated with various diseases?

8 Thank you.

9 DR. REEDE: Thank you very much.

10 DR. BOUGHMAN: It is because of this change in science in general that  
11 the American Society of Human Genetics has been asking the question, as if we didn't have  
12 enough questions at the professional level and the health professional level, we have asked the  
13 questions kind of on the front end.

14 How can we work together better to improve on our answers to several of  
15 these questions? We asked ourselves, what resources do we have, and I'm speaking now as the  
16 umbrella organization of genetics, knowing that the American College of Medical Genetics,  
17 and the other medical genetics organizations would be here and be able to say some of the same  
18 things slightly more focused, but I'll speak in the broader terms.

19 What we bring to the table, if you will, is human genetics resources, in the  
20 form of individuals who have a knowledge base. What we have with the other organizations  
21 here, and as Dr. Dunston has pointed out, in the general public is a set of needs and questions  
22 that are being asked. So it seems to me, anyway, that we are now at almost the point of what I  
23 call genetics readiness across the board.

24 People are beginning to identify genetics as interesting, as important, it is  
25 appearing in the newspaper and so on, and now they want to ask the more practical question.  
26 We, the geneticists, are saying we have some information, and it is the gap in between. The  
27 geneticists need to become more teaching ready, and the organizations and communities need  
28 to tell us more specifically what they need.

29 At that interface, I believe is the concept of models or materials, as Joe  
30 McInerney pointed out before. And now we have had two tries at DNA Day, and each one has  
31 been better than the former one. This year, we got a couple of very interesting responses to our  
32 survey, and some terrific ideas on how to engage second and third graders in some movement,  
33 and some cutting and pasting kinds of activities that talk about differences and similarities, and  
34 so on, in a very practical kind of way. So some new kinds of things that we think can be put  
35 out there.

36 Exactly the same thing would apply in public health in our work with the  
37 American Academy of Family Practice in their genetics initiative and so on where the needs  
38 and the human resources are coming together in the form of models to tell us how we can be  
39 more helpful in the development of certain models, although we don't have the expertise to  
40 actually translate and put those onto the  
41 CD-ROMs. We are going to need the resources together to get that activity and bring the other  
42 professionals in that need to focus on that. But I think we have the major portions covered, and  
43 now we just need the impetus to put it together and build the rest of the model together.

44 DR. REEDE: Thank you.

45 Ms. Allain?

1 MS. ALLAIN: Jo actually very eloquently said much of what I think  
2 NSGC's perspective would be on this. I think we are bringing to the table the human resources,  
3 we are at a number of tables now, along with ASHG and NCHPEG, and other organizations.  
4 Primarily NSGC has been focusing on in the last year, the allied health professions, by joining  
5 the Health Profession Network, which is an organization of allied health provider professional  
6 organizations, as well as joining the Association of Schools of Allied Health Providers. So  
7 trying to be the resource to integrate genetics curriculum into allied health provider education,  
8 as well as ongoing education for those professionals.

9 But as Jo pointed out, we have the resources, they have the interest, but it is  
10 getting the  
11 evidence-based activities, developing case studies based around those specific disciplines, in  
12 order to integrate those into their curriculums and daily activities.

13 DR. REEDE: Mr. Citrin?

14 MR. CITRIN: For the schools of public health, I should note in terms of  
15 your question of what actions are necessary in order to train faculty, I should note the role that  
16 the Centers for Disease Control and Prevention has been playing in moving genomics more into  
17 the forefront of both research, particularly teaching, and practice connections with the schools  
18 of public health.

19 The Center, and of course, Dr. Khoury, is the resident expert on this  
20 program, and your committee, and nationally, has been funding three centers for genomics and  
21 public health, of which Michigan is one, and North Carolina and Washington are the other two.  
22 Part of our role has been, up to now, to advance knowledge of genomics in public health, and  
23 to further the interaction between the schools and the world of public health practice around  
24 genomics, and the integration of genomics into public health practice.

25 I should say one of the facilitating factors here is that at the same time that  
26 the schools of public health are increasingly trying to develop closer relationships with public  
27 health practice, genomics being in an early stage of expanding in public health practice,  
28 presents a wonderful model for the schools on how to connect with practice. Because the  
29 schools and practitioners can be learning about integration and expanding knowledge in the  
30 field at the same time, and can be keeping pace with each other, and performing the usual loop  
31 of research applied into practice, practice being studied by researchers. Genomics is just a  
32 wonderful model of how to do that.

33 It seems to me that the associations, and here in our case, our own  
34 association of schools of public health, can do a lot to facilitate the linking up of people within  
35 each of the schools who are carrying out these roles within the schools of expanding the  
36 teaching of genomics.

37 CDC and their centers program have certainly been supporting this, and  
38 they are about to expand their program of centers. They may not all be in schools of public  
39 health, but they're all going to be involved in public health teaching and practice, and sharing  
40 education.

41 All of what has been said about the increasing resources that are available  
42 for teaching needs to be paralleled, it seems to me, by advocates within the institutions of  
43 learning who not only carry out the roles of spreading out the need for teaching in their own  
44 schools, but need to be able to connect with each other. Here is where the associations can do  
45 an awful lot to form committees that connect people across, in our case, the schools of public



1 health who are trying to advance teaching of genetics in their schools, so they can be sharing  
2 lessons learned, the barriers, and how to overcome them.

3 DR. REEDE: Thank you.

4 I'd like to open the questions up to the committee now.

5 MS. ZELLMER: Does anyone have any comments on ways that  
6 practitioners, not students, that we can get the word out to them? I mean, I'm the parent of a  
7 child with a genetic disorder, and the frustrations in dealing with medical professionals, it  
8 seems like medical students, nursing students, you've got a captive audience, and that's  
9 probably the easier question.

10 You can teach them, and my experience was I've got a husband who is a  
11 physician, and I have got a father who is a physician. We saw four different physicians. The  
12 first time we were recommended genetic testing was by a resident. I think the problem I see is  
13 how do you get the people that are already out there practicing to realize the importance of  
14 genetics?

15 At least I would guess with continuing education, it is strictly voluntary. I  
16 mean, they are going to go to the things that they want to go to, and I think probably most  
17 physicians don't understand the importance of genetics. How do you encourage them, or force  
18 them to learn what they need to know about genetics?

19 It seems we have the materials, but how do you get them to use them?

20 DR. REEDE: Dr. Lashley?

21 DR. LASHLEY: I would make a comment in regards to nursing in that  
22 regard. In some states, continuing education is in fact a part of relicensure, or for  
23 recertification for specialists, depending on your specialty. But I think the aspect that we  
24 briefly touched on before, which is requiring a certain amount of genetic knowledge, much in  
25 the same way that HIV knowledge is part of mandated, continuing education in many states  
26 would be one way to unfortunately, one has to look at driving curriculum sometimes by  
27 licensure and certification aspects. That is one way to do it.

28 Another way is to go through the specialty of continuing organization's  
29 genetics in each of their programs, whether it be as a feature by itself, or integrated within such  
30 things as MI, if it is a cardiology conference, updates in that, or whichever way that goes.

31 DR. REEDE: I'd just like to acknowledge that Dr. Reed Tuckson, a member  
32 of the committee, has now joined us. Welcome, Reed.

33 DR. TUCKSON: I'm listening carefully, and I will chime in if I get a  
34 chance. Thank you.

35 DR. REEDE: Dr. Boughman?

36 DR. BOUGHMAN: Let me put on a slightly different hat for just a  
37 moment. For the last 14 years, I have been a representative from the American Board of  
38 Medical Genetics to the assembly of the American Board of Medical Specialties. There have  
39 been two or three things that have happened in the last five years or so in that organization.  
40 Coming back to the comment that Dr. Lewis made earlier about statements and accrediting  
41 bodies being extremely important.

42 One of the things that has happened is the shift in continuing medical  
43 education to required maintenance of certification. This is a formal shift, this is a difficult shift  
44 for many of the specialties to make that once had the attitude, once I pass my boards, that's  
45 really it.

1                   So we do have a window of opportunity here to in fact include genetics as at  
2 least recognizably new information. Our challenge is to make it the most exciting among the  
3 various kinds of continuing medical education courses, or whatever, that individuals can take.  
4 So it is a two-way street there, certainly.

5                   One of the other comments that I would like to make is that as the genetics  
6 representative to the process of creating the principles of practice in training and in  
7 maintenance of certification, I think it was the approach of the geneticist and our tradition in  
8 number one, looking at family units, and number two, fully recognizing and addressing  
9 cultural-specific issues and the need for cultural competence in every clinical situation.

10                  I think our experience in that helped bring that to the fore, and I think that  
11 may be the results of the way, if you will, geneticists kind of look at the world at the same time  
12 we're looking at individual patients. But we do have a window of opportunity here in  
13 maintenance and certification for physicians.

14                  DR. REEDE: Dr. Lewis was next.

15                  DR. LEWIS: One of the things that we've been working at really hard with  
16 ISONG is to make people with specialized knowledge available for professional meetings. For  
17 example, I'm speaking with three colleagues, one of whom I believe became interested in  
18 genetics because she has a child with a genetic condition, and the nurse from NIH who is her  
19 research nurse, and another colleague and myself, the four of us are presenting at the American  
20 Nurse Association, which is probably the biggest generalist nursing organization.

21                  Dr. Lashley is presenting there too this year, and so we're working really  
22 hard to present at non-genetics conferences, and to be able to work with our colleagues. Now  
23 obviously when I'm speaking, I'm one of 27 break-out sessions, and how many people are going  
24 to choose to go there versus retirement planning, versus how to take care of the patient in the  
25 intensive care unit? That becomes a hard issue.

26                  I think part of what happens is that as people are exposed and learn from  
27 people like you, because I learn most of what I learn not just from books, but from the patients  
28 that I see every day. So as I interact with people like you and your family, that hopefully will  
29 pique my interest in that this is an area where I need more education. So when I go to my  
30 professional meeting, I'm going to choose the genetic session over the retirement planning  
31 session, or whatever.

32                  So that I think it is a two-way street. But to forget that patients are probably  
33 our most valuable source of education, and that is not helpful as a patient when you're going in  
34 to realize that you're as responsible for educating the clinician as the clinician. So if you look  
35 at it as a partnership, I think sometimes that becomes real helpful, too, and that each of us has  
36 an opportunity to help educate folks.

37                  I'm sure that the folks that you and your family dealt with learned as much  
38 as you did, it sounds like, but that becomes an important partnership, I think, too.

39                  DR. REEDE: Mr. McInerney?

40                  MR. McINERNEY: Yes, thank you. Professional societies are an  
41 extremely important vehicle for reaching out to health professionals. I'll give you four concrete  
42 examples, one of which Ms. Allain can describe better than I. But the National Society of  
43 Genetic Counselors worked a couple of years ago with the Endocrinology Society to integrate  
44 genetics thoroughly into its annual meeting. I don't remember how many members of NSGC  
45 were there, but I think there were at least 20 members of the society who were at that meeting.

1                   They actually constructed genetic counseling sessions around a particular  
2 genetic test for the endocrinologists, so they could come in and do hypothetical genetic  
3 counseling sessions. The point here, and you'll hear it again, is to work with the professional  
4 societies so that you are meeting their needs.

5                   NCHPEG will be working with the National Black Nurses Association to  
6 develop a half-day workshop on genetics for its annual meeting in 2005. Beginning this year in  
7 October with its annual meeting in Orlando, the American Academy of Family Physicians will  
8 begin a  
9 year-long clinical focus on genetics, and there are lots of organizations involved in this.

10                  The Genome Institute, I think CDC is involved in one way or another, and  
11 that will continue for the year. But again, we worked with the society, the Academy, to  
12 determine what their needs were. I thought it was very interesting, when the group of  
13 geneticists sat down to develop 12 modules, one per month for the clinical focus, when we  
14 looked at it, it was what geneticists would come up with.

15                  When we looked at what the interest of the family physicians were, there  
16 was almost no connection, except for the fact that each of the diseases, and they were all  
17 disease oriented, each of the diseases has a genetic component. But the importance is to work  
18 with the professional societies to build those programs. So there will now be one module a  
19 month rolled out by the American Academy of Family Physicians in conjunction with a number  
20 of us around this table, to bring genetics to family physicians once a month over the year.

21                  We have also just finished working with the American College of Clinical  
22 Pharmacy to develop a new continuing education program on pharmacogenomics. And again,  
23 they came to us as a genetics community, and we have been working with them now to bring  
24 the genetics that they need to that program for their members. So professional societies are  
25 extremely important.

26                  DR. REEDE: Thank you very much.

27                  I have a general question. As I'm hearing about these various forms of  
28 curricula, the cases, the different materials that are being developed, is there a mechanism in  
29 which those can be shared? Part of my concern is everyone is starting from scratch every time.

30                  MR. McINERNEY: If I may, that is why NCHPEG exists. If you come to  
31 our website, you'll see a list now of about 55 different educational resources in our database,  
32 about ten of which we've had reviewed. Two reviews usually, one by a member of the  
33 profession for whom the materials are intended, and the other review by a practicing geneticist.

34                  There are also lots of other resources on our website, but that's why we  
35 exist. If you have materials that you would like to bring to the notice of the community, please  
36 let us know, and we'll link to them, or we'll put them on our website.

37                  DR. REEDE: Thank you.

38                  Dr. Felix-Aaron, you had a question?

39                  DR. FELIX-AARON: I didn't have a question, I had more a comment on  
40 what Kimberly said in terms of trying to reach practicing providers. Somebody said earlier in  
41 terms of the window being open, I think that is right. But I think in addition to the window  
42 being open, sort of being strategic about the types of providers you engage, would be something  
43 that would be important for this group to consider, if indeed that was the direction that it  
44 wanted to go forward.

45                  So I think it is important that the family physicians are coming to this issue,

1 and they have that particular focus. But I think there is also a role for the committee in terms of  
2 understanding the science, and understanding the progression of the science, and with the  
3 progression of the science, which types of providers, whether they be physicians or nurses, are  
4 at the forefront of the evolution of that science? Which organizations we need to target as a  
5 committee, and which professional groups that need to be targeted as a committee.

6 So just to summarize my main point, is that clearly there is a need for  
7 targeting in terms of the providers that are on the forefront. I would imagine that the cardiology  
8 is important to engage the cardiologist, that the pediatricians, I mean, the state of the science is  
9 such that engaging the pediatric community would be much more of a priority than say  
10 engaging someone with subspecialties.

11 DR. REEDE: Thank you.

12 Dr. Leonard?

13 DR. LEONARD: So on a national level, CME is, I think, directed on a state  
14 level. Is there a national mechanism to mandate CME in, or continuing education? I call it  
15 CME because I happen to be a physician, but continuing education, professional education, for  
16 all groups at a national level, rather than doing it state by state, or organization by organization?

17 DR. REEDE: Dr. Whitcomb?

18 DR. WHITCOMB: The simple answer is no. Licensure is granted by states  
19 in medicine. As a matter of fact, not all states even have requirements for any continuing  
20 medical education. About one-third of the licensing authorities do not, and of the states that do  
21 have requirements for licensure, or relicensure I guess I should say, very few of the states  
22 mandate any specific content that needs to be covered as a part of the relicensure.

23 There are some that do, but as a general rule, the answer is no. But if one  
24 wanted to think about this across the country as a whole, the answer is there is no mechanism in  
25 place to accomplish that.

26 DR. REEDE: Brad?

27 MR. MARGUS: Thanks. So the people in this room obviously, I think, all  
28 concur that genetic education is important. I feel a little bit like in several other meetings, too,  
29 everyone is preaching to the choir, there are all these reasons why science is changing, and  
30 people need to be kept up to date and all that.

31 I'm a little unclear about what genetic education means. If it is how genetics  
32 works, so that a physician, nurse, or counselor has to be able to explain Mendelian inheritance,  
33 or what the risks are of a particular test they're going to have, or if they are supposed to, as in  
34 Kimberly's case, know the catalog of all the rare disorders and be able to help in diagnosis, that  
35 is a completely different and much more challenging thing. Or is it the ELSI part that we  
36 always hear about?

37 But either way, it is pretty clear, and our resolution is going to say that  
38 education is really necessary, it has to be coordinated, and all that. I think, though, because if  
39 this committee is really going to have any impact, it is kind of like the Genetic Discrimination  
40 Act, which is we're going to have to tell the Secretary that something is definitely broken.

41 I'm not really hearing that something is broken, so maybe things aren't  
42 broken, and certainly if you look at diversity among genetic counselors and things like that, we  
43 can go down there. But is there something that you can point to that it is really broken? Can  
44 you bring a large number of patients or families in front of us, or in front of the world and say,  
45 these patients were mistreated because their physician was clueless about genetics, or got it

1 wrong?

2 I haven't heard any of those stories. I have only heard that kind of like we're  
3 anticipating that this is all coming, and we'd better be ready, and we only have one genetic  
4 counselor for millions of potential patients who are all going to be screwed up if we don't have  
5 more educated people out there. But today, is anything really broken? If it isn't, it is going to  
6 be challenging to get more funds in the things that we're asking for?

7 So if the people who are going to continue to make comments could just  
8 highlight for me anything you can point to that is really, really broken, it would be really  
9 helpful. Again, if part of the genetic education and continued education is things like knowing  
10 everything about rare diseases, I know from my experience as an advocate, I could bring  
11 forward thousands of families who, like my family, spent a year and \$60,000 of needless tests  
12 to find out what the real disease was that my kids had, because the physicians at world class  
13 medical centers and the geneticists didn't think of the right disease to test for.

14 But other than that, other than knowing that kind of information, which I'm  
15 afraid maybe it is always going to be a challenge with rare disorders, all the other things that  
16 we're talking about is part of genetic education. Is it so bad right now that things are really  
17 broken?

18 MS. ZELLMER: I just want to make one comment, though, Brad. I  
19 certainly don't expect every physician to know every rare disorder, and I think you're right, that  
20 would just be a useless resource. What I was looking at more is just the fact that a physician  
21 should recognize that there could potentially be some genetic basis for the problems that the  
22 child is having.

23 Certainly my child's disorder is rare, but if you look at the symptoms that  
24 she was having, in looking back on it now, I think it should have been something very obvious  
25 to a physician that genetic testing should be recommended to us, that there were potentially  
26 genetic bases for the problems, and certainly not that she had this specific disorder.

27 I think that it was very clear to a resident who saw her for one time, well,  
28 you know, have you ever had genetic testing? I think something is broken when you have  
29 several doctors, and it takes them two years to even recommend genetic testing for a potential  
30 rare disorder. Not that they know the specific rare disorder, but that genetic testing is even  
31 recommended as an option. That would be my recommendation. I think something is broken if  
32 it takes two years to even have genetic testing recommended.

33 DR. REEDE: Mr. Citrin?

34 MR. CITRIN: Let me suggest, and I'm delighted that question got put on  
35 the table, because one of the comments when we got around to any specific comments on the  
36 draft resolution, one of the comments that I was going to make was that the resolution could  
37 carry more of a sense of urgency to get on with this activity.

38 Let me suggest three areas where it is not so much that things are broken,  
39 but that things are, at this point, moving towards what could be very significant crashes. One is  
40 in the area of costs, that when you look, for instance, at genetic testing, here is an expanding  
41 technology as to which the corporate sector, the  
42 profit-making sector has an incentive to get more and more tests used, far beyond what the  
43 scientific sector is able to demonstrate is effective, valid, and cost-effective.

44 So the ability to teach people who are professionals on how to assess and  
45 evaluate the worthwhileness, the validity of this expanding technology, is absolutely critical. If

1 people, whether they are medical professionals or public health professionals, don't have this  
2 ability to assess the worthwhileness of this vast array of exploding technology, what we'll end  
3 up with is an enormous wasteful use of health care resources.

4           Secondly, there is this whole matter of distortions that are occurring in the  
5 public's view of genetics, as the result of where the public is getting most of its information  
6 from. The public, of course, is getting most of its information on genetics, as well as it does  
7 other subjects, from TV, from media generally, relatively little from health care professionals,  
8 and almost none from public health.

9           The messages coming to the public now are both deterministic in terms of  
10 media hype on genetics controlling her conditions, which it really doesn't, as well as an item  
11 which is on your agenda, fortunately, and that is the direct-to-consumer advertising, which will  
12 further confound the public's view of what it means to have this or that gene.

13           And so here again, it seems to me we need a cadre, an increasing cadre of  
14 professionals who understand how to inform the public about genetics. This is a particularly  
15 critical role for public health people, because we see ourselves as having a role of public  
16 education, and here again, this happens to be one of the major things that CDC has been  
17 emphasizing in their new announcement of funding of genomics centers, this role in trying to  
18 correct distortions in the public's mind.

19           The third area where we could be heading toward a serious crash is one that  
20 is directly relevant to what Dr. Dunston, at my left, was talking about. That is the fact that we  
21 have here an expanding science that has the potential to exacerbate health disparities, at the  
22 same time that it has the potential of reducing health disparities. It is uncertain at this point as  
23 to which direction it is going to travel.

24           How our professionals learn about genetics, how they use that learning  
25 informing practitioners how to practice genetics, and how, again, they inform the public about  
26 what is the meaning of genomics, will have a lot to do with whether this new technology is  
27 going to create further gaps between the haves and have nots, further stigmatization of groups  
28 that have already been stigmatized too much, or whether genomics as a new, powerful tool to  
29 address some of the diseases that are responsible for which genetics has a component, and  
30 diseases that are responsible for health disparities, whether this very powerful tool will be used  
31 to reduce disparities.

32           So this, it seems to me, would suggest at least three areas where action is  
33 absolutely essential if we are not going to see this science lead to crashes in the future.

34           DR. REEDE: Dr. McCabe?

35           DR. McCABE: I want to make several comments about several of these  
36 points. First is one that I don't think I have heard here, and if I did, I missed it. That is in  
37 preprofessional education, and I'll focus on premedical education, since that is what I have  
38 more experience with, and the whole concept that we should really equip our professional  
39 students when they arrive in professional school with some background in this area.

40           The topic that I find particularly appalling in premedical education is the  
41 fact that we still require organic chemistry. I quite honestly have not formulated a medication  
42 once in my career. It is simply there as an energy barrier to premedical students, and yet,  
43 something as important as genetics, or one could pick another topic that has relevance to 21st  
44 Century medicine, is not required.

45           I would urge that in each of these areas, that we begin to look at what the

1 prerequisites are, so that our students come understanding the importance of these areas. With  
2 respect to the integration of content that was mentioned, I think it is important to note that some  
3 of our revamping of medical school curricula are treating genetics as a threat that runs  
4 throughout the medical curriculum, not as a specific topic.

5 I think it is important that we make it fundamental and exciting, and case-  
6 based on all of those topics. The degree with which we are successful does have to do with the  
7 politics of curricula, and arguments about crowding of the curriculum. So again, to the extent  
8 that organizations such as the AAMC, or other professional organizations, can encourage that  
9 this be a part of the curriculum, that will help those of us who do have to deal with the realities  
10 of fighting for space in the curriculum.

11 I think it is also important to note the cost benefits of genetic education. It  
12 does prevent the diagnostic odysseys that have been mentioned, which in fact are cost-  
13 beneficial. Off site, several examples with which I'm familiar where these diagnostic odysseys  
14 have occurred with delay and treatment of management of patients.

15 PKU, the classical newborn screening test, screening of screening, not every  
16 child will be identified, and those that are missed frequently go on for years before somebody  
17 suggests that the appropriate test be done. There are numerous examples of  
18 hemoglobinopathies, sickle cell disease, and thalassemia.

19 Again, relatively common disorders, and certainly quite common in specific  
20 communities in this country, that are not understood really by practitioners. Cystic fibrosis is  
21 another case where individuals can go on for years and years before they are identified.

22 Finally, we need to make sure that all of our professional students  
23 understand that this knowledge will prevent medical/legal mistakes, and medical  
24 misadventures. There is a big focus on medical errors these days. We talk about drug errors  
25 and all of those things, but errors of omission, errors of lack of diagnosis, or misdiagnosis, are  
26 also extremely important, and we have to recognize that not only are those cost inefficient for  
27 the families, they lead to tragedies where diagnoses are not made, or made too late. But  
28 ultimately, they also cost society, both in productivity, as well as in real dollars for those  
29 diagnostic odysseys.

30 So I think that there are a number of ways that we can approach this. If we  
31 don't do this in the health care professions, then our colleagues, some of whom are sitting  
32 around this table in the legal profession, will force us to bring this to the fore in health  
33 profession education. They will point out the misadventures, and the costs of those  
34 misadventures will become even higher with medical/legal actions.

35 Thank you.

36 DR. TUCKSON: Can I get put on the list, also? This is Reed. Thanks.

37 DR. REEDE: Reed? Go ahead.

38 DR. TUCKSON: Oh, okay. One of the things I also found valuable, the  
39 phrasing of the question -- can you hear me okay?

40 DR. REEDE: Yes, we can.

41 DR. TUCKSON: I thought it was valuable, the phrasing of the question  
42 around what was the problem, what was broken, what do we fix? I think two things. One,  
43 based on what Ed McCabe just said, that the Secretary of Health has a new initiative around  
44 health information infrastructures. I would wonder whether or not we might be able to have  
45 some relationship, or propose some initiative with that new health information infrastructure

1 task force to find ways in which we can support the availability of the best evidence-based  
2 science, as it is continuously updated, in an easy and accessible way for a clinician.

3 Obviously, nobody could possibly keep up with all of those permutations of  
4 information and knowledge in this field. So what I'm hoping is that somehow, one of the  
5 solutions may be that we could find a way for the Secretary to use this information task force as  
6 a potential solution to the availability at the point of care for people as these information  
7 systems are standardized, for both the outpatient, as well as the inpatient environment. That  
8 might be an idea.

9 Secondly, I think that the point made earlier about the maintenance of  
10 certification is exceedingly important, because at the end of the day, it is what you hold  
11 clinicians accountable for in terms of their criteria for certification. I think they, like the MS, is  
12 an essential group of people that we need to bring in.

13 Lastly, I would hope that maybe we would discuss later in this meeting,  
14 some things on HRSA. We will get to the idea of performance assessment. When you hold  
15 people accountable for their performance, that becomes perhaps that national unifying standard  
16 that someone asked earlier about, for getting their attention. So that as we start to get to  
17 performance assessment and it starts to get into the area of the use of technologies for genetics,  
18 then I think you provide a fertile environment for people to want to access the best appropriate  
19 evidence-based information, and then apply it. That provides, I think, a stimulus to go forward.

20 Thank you.

21 DR. REEDE: Thank you very much, Reed.

22 Sarah, do you have some comments on the committee that Reed was  
23 referring to?

24 MS. CARR: It's just to point out that this would be a very opportune time  
25 to make a suggestion to the Secretary about that. The President has actually asked for a  
26 strategic plan on the use of the improved use of health information technologies. So it would  
27 be an opportune time.

28 I do think that the more specific the committee can be about how that  
29 technology and genetics would dovetail, I think the better. That would be very helpful to the  
30 Secretary, I think.

31 DR. REEDE: Thank you.

32 Ms. Berry?

33 MS. BERRY: I'm not trying to stir anything up here, but I was curious in  
34 reading the different comments on this issue of education and training of health professionals, I  
35 was wondering whether there is any disagreement about who does what?

36 The reason it is a concern to me is because in the area of coverage and  
37 reimbursement, which we're going to talk about later, I know who does what will have some  
38 impact on whether an insurer, or whether a federal health program, will cover and reimburse for  
39 a particular service.

40 So I didn't know if we needed to go down this path or not, or whether  
41 everyone just prefers that we ignore it and have everyone sort it out amongst themselves. But  
42 that was an issue that popped into my mind, and I would love to hear your thoughts.

43 DR. LEWIS: I just want to speak briefly. Back before when we were  
44 talking about credentialing, and you basically said there were no national standards in  
45 medicine, that's not necessarily true in all of the disciplines.



1 Any advanced practice nurse is required to recertify every three years, and  
2 the recertification usually has both an educational and a clinical practice component. While I  
3 may not be required to do mandatory continuing education for the State of Virginia to keep my  
4 license, one of the things I have to do as an advanced practice nurse is I have to maintain  
5 credentials as a certified nurse, and that requires me to have 45 hours of continuing education  
6 every three years, and a practice component that is at least equivalent to a half a day a week of  
7 patient care, depending on my particular discipline.

8 Some states, Massachusetts, for example, requires 15 hours every three  
9 years for all nurses to maintain their licenses. So I think that there is variability among the  
10 professions, but to look at the fact that there are windows of opportunities, at least for some  
11 disciplines to start to look at the fact at, at this point, for example, as a women's health nurse  
12 practitioner, of my 45 hours, 30 of those have to be core, and 15 of those have to be supportive,  
13 but 30 of the 45 hours have to be clinically based.

14 So that there are opportunities in some disciplines to produce programs that  
15 would be attractive to people, and have them out there, and it may well be variable. Even  
16 though people are licensed by the state, in nursing, the credentialing is national.

17 DR. REEDE: Thank you very much.

18 Dr. Khoury?

19 DR. KHOURY: Thank you very much for all your presentations this  
20 morning. I may be a bit out of sequence, I have been gathering my thoughts for a little while  
21 here. I have a couple of comments, and maybe a question of the group.

22 We all talk about education and training, and I think many of the issues I  
23 heard this morning are right on target. There is sort of a two-prong approach to this, at least in  
24 my own mind, and I may be saying those same things tomorrow morning when I talk about the  
25 public health approach to genomics.

26 The first thing to keep in mind is the issue of genetic diseases, and the rare  
27 conditions that individually may be rare, but in aggregate, may affect about 5 to 10 percent of  
28 the population. We have heard about the diagnostic odysseys that cost money, cause anxiety in  
29 families, and destruction of the social and familial fabric, but also sometimes leads to medical  
30 issues.

31 Dr. McCabe mentioned cystic fibrosis, and I can count a number of other  
32 conditions where there are interventions. One example comes to mind, that is familial  
33 hypercholesterolemia, which is an LDL receptor defect that leads to premature heart disease,  
34 and people die in their 30s and 40s from heart attacks. That is about 1 in 500 disease, about a  
35 million people in this country may have familial hypercholesterolemia, and there are data from  
36 population surveys that about half or more of these patients are missed by the medical system,  
37 because there are so many people with high cholesterol levels due to other more polygenic or  
38 multifactorial causes of high cholesterol levels, that people with LDL receptor defects may be  
39 missed completely.

40 So if not for anything else, we need the kind of red flag raising. We are not  
41 trying to get everyone to become a geneticist obviously, but it sort of raises a red flag so that  
42 appropriate follow up can be made.

43 But coming back to what I heard also throughout the committee, and I think  
44 Dr. Whitcomb and others mentioned the issue of relevance today. Because for most  
45 practitioners, these kinds of incidents may be too far and too few in-between, I guess, they may

1 not be seen by one single practitioner on any given day, week, or month. So what else is going  
2 on? I want to paraphrase Judith Lewis' comment about the fever.

3 What is the equivalent of fever, a febrile episode in genetics? I mean, you  
4 made the analogy of the infectious disease specialist, so okay, you've got the patient with fever,  
5 and you're not going to refer everyone who shows up with a fever to an infectious disease  
6 specialist. Fever is a very common occurrence in the population. So what is the equivalent of  
7 that in genetics? To me, the equivalent of that in genetics is the occurrence of something in  
8 your family.

9 We have plenty of data that shows that at least the major killers, heart  
10 disease, diabetes, and cancer run in families. If you have at least one affected relative, you are  
11 at increased risk of these conditions, and they may not be due to a single-gene disease, and you  
12 may not have to refer every single patient with a family history to a geneticist. And yet, it is the  
13 equivalent of a fever, because half the population may have a family history of something, and  
14 that becomes sort of the run of the daily practitioner. Therefore, the relevance of  
15 genetics/genomics/family history becomes of urgency to the general practitioner, in addition to  
16 the issues of rare single-gene diseases.

17 I'll talk tomorrow about the public health approach to family history we  
18 started, but I'm curious to see what the various organizations are. I know some of them have  
19 done a number of things in this area. I'd like them to expand on the equivalent of fever in  
20 genetics, things like family history and the development of tools that everyone can use today,  
21 and we don't have to wait 10 years to show relevance of genetics in practice.

22 DR. REEDE: Would someone like to respond to that question? Mr.  
23 McInerney?

24 MR. McINERNEY: Yes. NCHPEG develops, three times each year, a  
25 family history newsletter that is devoted to exactly the issues that Dr. Khoury is referring to.  
26 This actually was a bit of a retrenchment for us. Originally, we were supposed to develop a  
27 generic family history tool for use by all health professionals, and it simply proved to be  
28 impossible with the time and the resources we had available, so we backed off on that a bit.  
29 Our family history working group spent a fair amount of time on it, but concluded that it was an  
30 impossible task.

31 What we do now, however, is develop this newsletter that we put online  
32 three times a year, devoted entirely to the issue of the family history in health care. It has  
33 articles about family history, but it also refers to tools that other people are developing, such as  
34 the tool being developed by the American Society, the Genetic Alliance, and NSGC, and the  
35 tool that CDC is working on now.

36 So in fact when we talk with our colleagues in the health professions, we  
37 refer to the family history as the first genetic test. It is inexpensive, and it is relatively easy to  
38 do. We are also building an extensive section on family history into our new CD-ROM on the  
39 genetics of common, chronic disease, which will be out later this year, and that is intended for  
40 primary care providers and public health professionals.

41 At some point, I would like to respond to Ms. Berry's question about is there  
42 an agreement about who does what, and who is going to pay for it. But I'll come back to that,  
43 perhaps.

44 DR. REEDE: Thank you.

45 Dr. McCabe?

1 DR. McCABE: Well, I just wanted to respond with an anecdote about the  
2 family history. In retooling our medical school curriculum, we had a laboratory in how to take,  
3 and how to interpret a family history. We were told by the organizers of the curriculum that  
4 that was not interesting, and was something that we really needed to completely rethink the  
5 next year.

6 On the other hand, I would point out that I'm Chair of a Pediatrics  
7 Department, I attend morning report whenever I can when I'm in town, and I'd like to think that  
8 our residents are more sophisticated in a family history now than a few years ago. They used to  
9 say, is there any family history? And now if one of them says that, the others will chime in  
10 with specific questions about what that is, rather than just do you have a family history of  
11 anything, which used to be the question, which the usual answer is no, or something completely  
12 irrelevant.

13 So I think that it is somewhat analogous to taking the temperature, but it  
14 requires a little more sophistication than simply taking a temperature. And yet, to many non-  
15 geneticists, it is considered boring, trivial, and uninteresting.

16 DR.. REEDE: Thank you.

17 Dr. Felix-Aaron?

18 DR. FELIX-AARON: I just wanted to elaborate a little bit on the  
19 Department's health information technology. AHRQ, the Agency for Healthcare Research and  
20 Quality, is leading part of this effort. We funded this year, \$60 million dollars in grants,  
21 specifically to do two things. One, to build the infrastructure on IT in small community  
22 hospitals, as well as in rural settings, and the other is to demonstrate the value of health  
23 information technology to health care.

24 So I think we have the opportunity to look at this really closely and to see  
25 where the opportunities are between what is going on in genetics/genomics, and what is going  
26 on in IT. If this committee was interested in that, I'd be happy to go back to AHRQ and look at  
27 the portfolios we have, and whether there were any grant projects that specifically looked at  
28 genomics, and I'd be happy to report to the committee on whether they were interesting  
29 programs, or interesting projects that was directly relevant to the work of this committee.

30 DR. REEDE: Thank you very much.

31 Dr. Guttmacher?

32 DR. GUTTMACHER: I'm just going to state the obvious. I think in terms  
33 of linking the number of the comments that we've heard, one, the range of comments about the  
34 importance of focusing on those areas of genomics that are actually usable and useful today to  
35 the provider. Two, this calls for thoughts about how we might feed into the Secretary's  
36 Committee on Health Information Infrastructure, and three, the importance of family history.

37 That really is the genetics tool that we can offer today that doesn't add cost,  
38 but in fact we think it is of some benefit, that reminds the practitioner of why genetic factor is  
39 important in health, and can be used as the basis for a lot of things. There is work that CDC is  
40 specifically doing, but others are doing as well to really further the idea of coming up with  
41 instruments and other kinds of things.

42 It seems to me it would be useful for the committee to call attention of the  
43 Secretary and his other committee to the potential use of family history, and the electronic  
44 medical record particularly. If we could establish the idea that everyone deserves a good,  
45 relevant family history and/or electronic medical record, we will have done a good thing for

1 patients, but we will also have established a template for providers to continue to think about  
2 other more sophisticated genetic testing and those kinds of things once they do become more  
3 broadly available and useful. So I would think that would be an obvious sort of first step to  
4 take.

5 DR. REEDE: Thank you very much.

6 Dr. Winn-Deen?

7 DR. WINN-DEEN: I think I greatly agree with the comments that Alan just  
8 made. I think one of the issues that keeps coming back to me about genetic testing, and I'm not  
9 talking about diagnostic genetic testing in the context of signs and symptoms, as much as  
10 presymptomatic testing, or maintenance of information about carrier status.

11 A lot of this information might be lost between the time a test is done, and  
12 the time the information is actually needed at some future date. I think it is very important that  
13 we, as we start to consider moving into the electronic age and doing electronic medical records,  
14 and having an IT infrastructure for that, that we have family history, we have a mechanism for  
15 recording any once in a lifetime genetic test that someone is given, so that that information is  
16 there. I'll say that extends to something as simple as blood type, and to things as complicated as  
17 pharmacogenetic drug metabolism enzymes that might be useful at multiple times in a person's  
18 lifetime.

19 So I would be very interested as a committee in hearing from this other  
20 group what they are doing, and how we could integrate these things together, because I think  
21 the combination of those two activities will help us to integrate genetics into the practice of  
22 medicine.

23 The third point that we have to lay on top of that is that there still are  
24 concerns, and Agnes mentioned this to me at the break, about having genetic information in  
25 your medical records, that some institutions still maintain your genetics record as a separate  
26 parallel file from your actual patient record.

27 So we need to deal with that issue. Why is that done still? Is it still an issue  
28 of fear of discrimination in some way in insurance, health, life, or employment? Or what is the  
29 reason that that is still happening? That is clearly a barrier to having a really fully integrated  
30 lifetime electronic medical record that can bring you the full benefits of a genetic test, rather  
31 than having to sort of hide that information, and then have it redone every time you need it.

32 So that won't deal with the situations where there is no family history, and  
33 we still have to train medical professionals to recognize in the absence of family history, which  
34 is what happens in a lot of rare genetic disorders, the probability of two carriers coming  
35 together is low, and so quite often they don't have any family history, but then they end up with  
36 affected children in recessive disorders. So I think both of those things are very important for  
37 us to keep in mind.

38 DR. REEDE: Just a question as you talk about this information, and are  
39 there questions with regard to confidentiality and sharing of information, it reminded me  
40 somewhat of some of the comments from Dr. Dunston, and the public's view of this  
41 information, how it gets shared, and concerns from special populations in terms of how this  
42 information gets shared. Could someone speak to that, please? Dr. Lewis?

43 DR. LEWIS: It's real interesting in terms of what people want in their  
44 medical records or not. I know when I deal with the women that I work with, for example, lots  
45 of folks don't want their sexual orientation in their official medical record.

1                   So I think when we start to look at information that is different information  
2 that is potentially labeling, it is genetics information, and it is a whole lot of other information,  
3 too. I'm not sure all of the privacy protections that we have in law that are so challenging to  
4 implement, necessarily help people in terms of the fear of what is in their record, or what is not  
5 in their record.

6                   I guess I worry about the fact that we have a system that has created the  
7 need for people to want information to be excluded from their medical record. I know several  
8 years ago when I went to Iceland and was sitting and talking to the President of Iceland about  
9 the issue of privacy and confidentiality, he basically looked at me and said, you know, that is  
10 not an issue in our country, because we are a democracy, and people trust the government.

11                   I thought, wow, how unique and how wonderful. For whatever reason, we  
12 have created a climate of distrust in this country. Not us sitting around the table, but the system  
13 has created a climate of distrust, and of people's need to keep private information private. So I  
14 think the problem is even more than genetic information, I think it is an information of the fact  
15 that people have been treated badly.

16                   Once people have been treated badly, or groups of people have been treated  
17 badly, then we have a lot of work to do, because once trust is lost, it becomes twice as hard to  
18 regain. So I do worry about people who have concerns about keeping information out of their  
19 medical record, and why.

20                   DR. REEDE: Thank you.

21                   Dr. Hans?

22                   DR. HANS: Thank you. I just wanted to extend some of the ideas that  
23 have been put forward around where this committee may want to consider interfacing with the  
24 Secretary's Health Information Technology Initiative. From the lessons of a department that  
25 has an integrated health information system, electronic health records available at the bedside  
26 for patients, some of the issues that you may want to consider along the following lines.

27                   One is certainly proposing what elements of genetic knowledge should be  
28 part of the basic components of the electronic health record. That is one of the decisions going  
29 on across the agencies at this time. Family histories could certainly be something that is  
30 considered, if you are out there as a technology developer, you should certainly put that  
31 capability into whatever system you are developing.

32                   I think it would also be helpful to look at the other kinds of tests and  
33 knowledge that should be part of the basic record, and part of the recommendations that this  
34 Department and others are looking at at this time.

35                   In addition, in a more future looking perspective, to think about once folks  
36 have records at the bedside and have the interactive capacity with those programs, what sorts of  
37 prompts and guidelines on a daily basis would be helpful for the practitioners gets a little bit at  
38 how much knowledge does every practitioner need to have?

39                   If you can build in some of that knowledge and reminder to the automated  
40 system, then you don't have to make all the effort to make sure that every practitioner knows  
41 every bit of information. But if certain diagnoses and conditions come up as they are put into  
42 the record, if there is an electronic prompt that then says what about this, or consult a genetics  
43 expert, you may want to consider this, that would be helpful in helping this entire enterprise  
44 around health information technology begin to map out where they may need to put that kind of  
45 information into the records over the next five to 10 years I think would be extremely helpful,

1 particularly from the professional societies.

2 Certainly that is the role many other professional societies are playing at  
3 this time in that arena, as providing information and guidelines that then can be incorporated  
4 into these electronics systems.

5 And finally, an area that I think both for genetics/genomics, as well as the  
6 rest of the fabulous knowledge that is coming out of NIH at this time is how do you incorporate  
7 knowledge management systems into a desktop electronic medical records system?

8 What do I mean by that? Well, you can't overwhelm a practitioner with all  
9 sorts of prompts and guidelines. We certainly found within the VA that after sort of a certain  
10 number of prompts, that physicians start to turn off the prompts. So in any one visit, you can't  
11 have too many things coming up on the screen at the same time. But you can think about a way  
12 where when you have a particularly difficult case and you're not able to diagnose what is wrong  
13 with the patient who is in front of you, if there is access on that desktop to information, ability  
14 to query based on the diagnosis in front of you, it can help physicians and other practitioners  
15 seek out information, and perhaps assist them in understanding sort of the conditions of the  
16 patient that is there in front of them.

17 So those are just some ideas that the committee may want to consider in this interface with the  
18 Secretary's committee.

19 DR. REEDE: Thank you very much.

20 Dr. Feetham?

21 DR. FEETHAM: Thank you. I have a number of things, and some of it  
22 goes back to the broader issues brought up in the original presentation, which I'd like to  
23 commend that you looked at what is common across the disciplines, because this is truly an  
24 interdisciplinary issue, and that we're really talking about quality of care and access. The way  
25 your report was framed, I think that comes forward as very important.

26 Many of the things that you brought forward are consistent with a report  
27 with recommendations we'll be taking to Dr. Duke, the head of HRSA, in just a few weeks,  
28 after doing an analysis across all of our agency about genetics activities.

29 But what I'm hearing is a key issue of communicating what is available to  
30 support the integration of the genetics to practice research and education. Again, part of what I  
31 see in the role of HRSA in working with our other federal and non-federal partners, is that we  
32 can facilitate, and we do have some mechanisms that we can help in moving forward with that.

33 Art even mentioned several activities, including we do fund the GeneTools  
34 with a contract with the University of Washington and Dr. Wylie Burke. This does emphasize  
35 current clinical labacobility as one example. Also, the Genetics in Primary Care Program we  
36 have had for several years is the focus of that, is getting that into practice, and I can give you  
37 several other examples. But the issue is how do we disseminate that? How does that get  
38 brought forward across all the disciplines, whether you are an active clinician or an educator? I  
39 think that is a key issue that perhaps this committee can look at in the future as an issue of the  
40 next steps of where we go.

41 Also in stepping back, some of the activities and current mechanisms we  
42 have within HRSA are we have programs that can look at cultural competency, and we have  
43 funding streams and programs in the Office of Minority Health, and other parts of HRSA, that  
44 that may be a help in what you're talking about.

45 We also have responsibility in workforce diversity, and have programs

1 within our Bureau of Health Professions that again, can be tapped into perhaps, and informed to  
2 move in this direction. Also we have programs looking at the pipeline for the workforce,  
3 whether it is a genetic specialist, or the generalist.

4 So again, just as a reminder, we do have some structures within our agency  
5 in working with our federal and non-federal partners that perhaps we can address some of the  
6 issues that you brought up in your original report.

7 We just had a meeting that Dr. Hans was at on Thursday, looking across  
8 information system capacity in our health centers. As you may know, we served 12.5 million  
9 patients in 3,500 sites in our federally-supported health centers, although we're only on average  
10 25 percent of the funding for the health centers. We can influence some of the direction, and  
11 one of the things we're working on within our health centers is the integration of the genomics  
12 into the latest science, which we do through our Health Disparity Collaboratives, which again,  
13 that has the patient registry with some of the prompts and activities that you have been talking  
14 about. So again, I think we can help facilitate in a variety of ways, some of the issues that you  
15 have brought forward with the review earlier.

16 DR. REEDE: Thank you very much.

17 Dr. Boughman?

18 DR. BOUGHMAN: I urge the committee in its deliberations to, and I'm  
19 quoting from a conference I was at recently, "don't let the perfect get in the way of good."

20 As far as Cynthia Berry's question on who is going to do this, and are  
21 people getting in each others' way, and are we stepping on toes. At this point in time, it seems  
22 to me that there is so much to be done, and there are in fact from the genetics point of view, so  
23 few of us to continue to do it, workforce issues being one of our issues, that from the  
24 educational point of view, that is not our primary issue.

25 It will be an issue if the question is, who is responsible for all this education  
26 without dedicated resources to get it accomplished? So as long as we can continue to generate  
27 possible resources from federal agencies, or from within institutions, then I think exactly who is  
28 going to do what is not our basic question. I think we have generated several models where  
29 we're working in concert with each other, and I hope that certainly continues.

30 The other point that I would make, and we have gotten some very  
31 sophisticated comments back about family history in the medical record, and specific prompts  
32 and all of those things, but I would simply remind folks that the approach that the Genetic  
33 Alliance, the National Society of Genetic Counselors, and the American Society of Human  
34 Genetics have taken in the development of a generic family tool as to remind people that it is  
35 the consumer's information, and it is the consumer's responsibility to gather that information to  
36 provide it to the provider, no matter who it is.

37 There is no blood test, there is no fingerprint, there is no way that any  
38 provider can get the information without engaging the consumer themselves. So in fact, the  
39 approach that we have kind of built our strategy on is let's get the consumer, let's get the general  
40 public excited about their own family history and information, some of which at least may be  
41 medically relevant. As they bring that to their provider, whatever the provider's credentials are,  
42 then in fact they will initiate and engage the provider.

43 If the provider, once again, has gone through their genetics readiness  
44 training, they will be much more able to accept and interpret that information correctly. But we  
45 have in fact not directly addressed this most perfect genetic or family history tool, and we are

1 working with other organizations to try and get down to the basics.

2 In many situations, we would take almost any information that we could get  
3 as a starting point, and I only need refer or remind people that many of the best genetic studies  
4 have gone back to family bibles, and the information that the family had to begin with to in fact  
5 garner our information, which would be considered the geneticist information.

6 DR. REEDE: Thank you very much.

7 Mr. McInerney, you had mentioned earlier wanting to make a comment in  
8 regards to who should be doing what. If you could speak to that, and also in some of your  
9 written comments with regards to the recommendations, you had mentioned promoting public  
10 education that provides knowledge and skills to consumers, with requirements to participate  
11 effectively with health professionals in decisions that informed genetic prospectus, which I  
12 think follows very nicely from the comments just made by Dr. Boughman.

13 If you could speak to both of those, please?

14 MR. McINERNEY: Yes, thank you. Well, Dr. Boughman really covered  
15 the first point that I was going to make, but I will tell you just a quick story. That is when we  
16 first published the core competencies in January of 2001, we heard a lot of complaints from  
17 people in the genetics community, that these competencies were trying to turn all health care  
18 professionals into geneticists.

19 I suspected that that was not likely to happen, to begin with. And in fact, it  
20 hasn't played out that way. I will say that the competencies are challenging, and we ask a lot of  
21 health professionals in those competencies. But what we've heard in the subsequent three and a  
22 half years roughly, is that health care professionals are quite sanguine about their own  
23 limitations, not only their own knowledge, but their own limitations in practice. They want to  
24 know how much they need to do to incorporate genetics effectively into their own practice, and  
25 when it is necessary, then hand that off to somebody else.

26 What we found is that people are using the competencies in that way. They  
27 are thinking carefully about how genetics manifests itself in their own practice, and then  
28 picking and choosing the competencies appropriately. So I think that issue has resolved itself,  
29 perhaps only for the moment, but I think it has resolved itself now. The issue of who pays for  
30 what, who delivers genetic services, that's an entirely different issue, and one that I'm not even  
31 remotely qualified to address. But from an educational standpoint, the other stories should  
32 serve, I think, as a helpful guide.

33 With respect to my proposed recommendation, or proposed addition to the  
34 recommendation you want to take to the Secretary, it just strikes me, as I said to the committee  
35 when I testified here at the last meeting, that we can't think about education of health  
36 professionals in the absence of education of the public, particularly if the assumption is that  
37 genetics ultimately is going to move health care more and more towards a prevention-based  
38 paradigm. That is something we have all been hoping for for a very long time, and for me, the  
39 notion of prevention always has implied a partnership between patient and provider.

40 If we are going to have an effective partnership, both of the partners have to  
41 be well educated. The education, I think, for the public, has to proceed from the same  
42 conceptual base, the same set of assumptions about genetics that we use for health care  
43 professionals. The details will differ, but I think the set of conceptual assumptions has to be  
44 congruent with that for health care professionals.

45 DR. REEDE: Thank you very much.



1 Dr. Dunston?

2 DR. DUNSTON: Yes. I wanted to underscore a point made earlier, too,  
3 about the importance of getting the consumer involved as part of this transition. But I wanted  
4 to preface my comments by saying that I thought it was very instructive that this committee is  
5 genetics, health, and society, and not genetics, disease, and medicine.

6 Instructive in the sense that I think that engagement of the consumer in why  
7 they need to be engaged in understanding the knowledge, participating in the research is a  
8 challenge that is part of what I like to call, the very positive potential of genomics. In that this  
9 seems to be a time where the genomic technologies offer an opportunity to understand disease  
10 at the biological level that requires the participation of the consumer, participation of the non-  
11 patient, and family histories must engage those who are not coming to the attention of the  
12 physician because of the disease.

13 The absolute necessity of public education, so that we can get participation  
14 in research to really utilize the power of genomics, and understand the biology of disease, is a  
15 challenge that I think is putting public health in the forefront. The necessity of the consumer,  
16 and the population, both those affected and non-affected with disease, are integral to  
17 understanding the biology of disease in ways that the challenge to me would be to take  
18 everything that we have learned on the negative side, if you will, from genetics with disease  
19 and medicine, and ensure that laws are put into place now that would at best, minimize the  
20 exploitative and manipulative, and the issues that would keep the public from participating in  
21 the research that is absolutely necessary to really use the power of genomics today to  
22 understand biology.

23 So on the one hand, we can take what we have learned during the era of  
24 genetics that may serve as the basis for the public being fearful, and look at the laws. I loved  
25 the beginning of this meeting with the emphasis on the Nondiscriminatory Act. We can ask,  
26 what are the things that were wrong with the way we used genetics before? And what do we  
27 need to put in place to minimize that occurring so that the science can move forward in a way  
28 that the public will want to participate?

29 I really underscore the importance of research that engages the public,  
30 engages the non-affected, as well as the affected as essential to getting the benefits of how we  
31 understand our genetics today.

32 I think, lastly, that for the public to understand that the power of genetics  
33 now, or genomics, large scale, high-throughput, information technology driven science, to tease  
34 out the underlying biology that is related to disease, requires public participation. So the  
35 question becomes what do we need to put in place with our laws, policies, and education, that  
36 will ensure that we do a PR job for what the potential of genomics is is one that we are not  
37 concerned about a whole battery of things to present discrimination. But what do we need to  
38 do to protect our privacy? To protect confidentiality?

39 Those are issues driven more by how do we get the public excited about the  
40 power of science today to help us understand disease in a way that gives us whole new  
41 approaches to prevention and promoting health.

42 DR. REEDE: Thank you.

43 Dr. McCabe?

44 DR. McCABE: I think that's a terribly important point, because if we are to  
45 really utilize the power of genetics, then we will need large population studies, and we'll be

1 talking about that more. But given that the unique genotypes, whole genome genotypes, that  
2 individual patients will have will be relatively rare, if we're really to develop the correlations,  
3 we will need those large population-based studies.

4 I think it is also important that you tie it to the Genetic Nondiscrimination  
5 Act that was discussed at the beginning of this by Dr. Boughman, because until our patients  
6 feel that they can be safe with genetic knowledge in their medical care, they're not going to feel  
7 safe in the research arena.

8 If they understand that the research is to improve their care, and not to be  
9 used in a discriminatory fashion, because that is outlawed, then I think that they will feel far  
10 more secure in gaining that knowledge about themselves. So I agree wholeheartedly with your  
11 points, Dr. Dunston.

12 DR. TUCKSON: This is Reed Tuckson. I'd like to get on the list also.

13 DR. REEDE: Reed, go ahead.

14 DR. TUCKSON: Thank you. Two things. I think it was very helpful to  
15 hear those last comments. One of the things I have been impressed by, particularly by certain  
16 people on our committee, and many of the people that have testified, is how specific they are of  
17 families who are going through the experience of genetic disorders and diseases, how quickly  
18 their learning curve reaches fairly impressive levels.

19 With that as a hopeful sign, I am also, though, faced every day with the  
20 reality of just how tough it is to get any level of understanding about health and disease in the  
21 general American population. If you even just take an example like obesity, it is a devil of a  
22 time trying to educate people in our country about the etiology and other therapeutic issues  
23 regarding something that is commonplace as that disease and disorder.

24 If you think even of the number of people who are currently appropriately  
25 getting diagnosed for hypertension, my point is that I think that while this is an important area  
26 that we have underscored, I think this committee is going to be challenged by trying to figure  
27 out what the priorities are in this area.

28 We obviously cannot reorder all science literacy in the country. In fact, if  
29 you look at the science literacy of the American population, it is staggeringly low. And so I  
30 wonder whether we might start to, as we continue to discuss, drill down to the key priority  
31 areas that we think we can meaningfully through the power of the Secretary, actually do  
32 something about.

33 Secondly and finally, I just wanted to briefly get at this idea of who does  
34 what, and what those competencies are that Joann talked about, and others. I hope that we can  
35 also get to this idea of defining what needs to be done, and then start to figure out the whole  
36 range of comprehensive tasks, and then start to determine what is the interrelationship between  
37 different parts of the health care system, and the various professional, and maybe even non-  
38 professional disciplines that have an opportunity then to play a role in that. I think if we talk  
39 about it, we can get more specific.

40 Thanks.

41 DR. REEDE: Thank you very much.

42 In the interest of time, if there are any comments or questions with regard to  
43 the recommendations for resolutions? Additions? Mr. Citrin?

44 MR. CITRIN: Well, the recommendations that I had as comments relate to  
45 a couple of my earlier comments, but just to be quick. One is that while public health is

1 mentioned in page two, there are references in page one to clinical medicine that could be  
2 broadened out to make clear that they relate to public health as well. There may be some more  
3 generic terminology. I still get the sense that there is more of a clinical/medical focus than  
4 there is a broader focus on all health professions and education, including public health.

5 Secondly, it seems to me that the ethical/legal/social implications, the ELSI,  
6 ought to be referred to specifically somewhere in the resolution. It is interesting that in the  
7 Institute of Medicine's report that I referred to on the teaching of public health, a good deal of  
8 the language in the recommendations on the genomic competencies that all public health  
9 professionals need, related to the ELSI dimension.

10 It isn't automatically evident in reading the resolution that this is a  
11 component of the education, that this committee particularly ought to see as essential since it is  
12 so much at the heart of what this committee is all about.

13 Then in terms of the preamble, and I guess this relates to the question that  
14 was put on the table earlier about what is broke, that it would be helpful, it seems to me, in  
15 gaining attention to the resolution if some content were put into the preamble to suggest some  
16 of the really compelling reasons why it is urgent that increased attention be given to the  
17 education of health professionals in genomics because of concerns of what will happen if the  
18 pace of development of technology goes far beyond the competence of professionals to make  
19 use of the technology, or to inform the public adequately about the implications of the  
20 technology.

21 Thank you.

22 DR. REEDE: Thank you.

23 Dr. Lewis?

24 DR. LEWIS: My only comment is on your points in terms of the  
25 recommendations on number five. I'm not sure who is going to decide what is adequate  
26 knowledge in human genetics and genomics, because that might depend on where you sit.

27 So to use something that is either to take out the adequate, or to determine  
28 how it is going to be measured would be helpful, because I think that having that be left to  
29 interpretation may be problematic.

30 DR. REEDE: Dr. McCabe?

31 DR. McCABE: I just wanted to follow up on Mr. Citrin's comments about  
32 the ELSI. Not only are they important, but also they are engaging to the students. Perhaps  
33 even more importantly, however, while the technologies will change, the ethical, legal, and  
34 social implications of those technologies will remain somewhat constant. So I think it is  
35 important to cast a lot of the teaching in those, because they are engaging, and they are  
36 important. But they also will be somewhat longer term issues than whatever is the technique de  
37 jure.

38 DR. LEONARD: This is a rather specific point, but in recommendation  
39 number five, "Encourage accrediting licensure and certification bodies to condition  
40 accreditation licensure and certification among demonstration of taking out adequate  
41 knowledge in human genetics and genomics." Can this be specified to refer to health care  
42 professionals?

43 Laboratories that do testing are also accredited and licensed, and I don't  
44 think you were referring to laboratory or testing accreditation and licensure. You are talking  
45 about accreditation and licensure of health care professionals. It raises a measure of concern

1 among those of us who do laboratory testing, because there aren't a lot of people out there who  
2 are accredited and licensed in genetics, such that if we are limited to hiring only people who are  
3 accredited and licensed, we won't have a workforce.

4 So while I agree that we should encourage accrediting licensure and  
5 certification of health professionals, it shouldn't include laboratories.

6 DR. REEDE: Any other comments? Dr.  
7 Felix-Aaron?

8 DR. FELIX-AARON: Yes, a couple of points that came up in the  
9 conversation today. I don't see them represented in the points in these documents. I want to  
10 build on Dr. Whitcomb's point, the question around building the evidence. So that not only  
11 relates to Dr. Whitcomb's point, but also to what Dr. Tuckson said about the what to do in  
12 terms of building the evidence base for genomics and its implication, not only for practitioners,  
13 but for medical education. So I don't see that reflected in the resolution.

14 The other point I would like to make is also related to this issue, but how to  
15 integrate content. I'm not sure what I'm about to say has implications. Well, it is something  
16 you would try to implement in this document, but I think it should be captured, and the  
17 committee may want to consider it in terms of how to integrate genomics into medical  
18 education.

19 I think we may want to study what HRSA has done with the disparities  
20 collaboratives, and how they have used a collaborative and improvement model to get changes  
21 in practices. So I offer this as a model that we could use, or at least study to see how you get  
22 change in medical curriculum. So it is an area which I think is right for a pilot.

23 You have best evidence, you have tools, you have people in small pockets  
24 doing best practices, and you could use that, and develop a model for getting that kind of  
25 curriculum changed into medical schools, schools of public health, and nursing schools. So I  
26 think it is something that I would like to put on the table that if the committee was interested in  
27 looking at.

28 It is something you could comment on sort of what the health centers have  
29 done, and how they have used a collaborative learning model, coupled with an improved model  
30 to change practice. I come from the Agency for Healthcare Research and Quality, and we think  
31 a lot about integration and trying to change practice.

32 We struggle with the issue of overburdening not only teachers and  
33 practitioners, but really trying to give people the tools, give them the tools, give them the  
34 setting they need to support and foster change in their practices.

35 DR. FEETHAM: I mainly would like to say I had written that down earlier  
36 that they, again, the collaborative or breakthrough series type of model may be something that,  
37 again, various disciplines could come together and make application to the Bureau of Health  
38 Professions to look at this as -- because system changes really are a major piece of what we're  
39 talking about. Not only within the academic institutions, but in the practice and providers.

40 So again, I would just encourage our community to be thinking of  
41 something innovative that you might look at that type of a model for coming in for funding  
42 from the various federal agencies to build off of that idea. Again, I can give people information  
43 about our website if you want to know more about the breakthrough series, and that type of  
44 learning model. But I do think we need to look at this within the context of system change.

45 DR. REEDE: Mr. Gray?

1 MR. GRAY: Yes, thank you. I'm sitting in for Commissioner Miller, who  
2 wanted me just to raise this one point, and I guess it relates to point number six. That is that  
3 we're talking about issues relating to diversity that we keep in mind diversity within the  
4 disability community. Cultural diversity should include that segment of the population as well,  
5 and we should just bear in mind that within the disability community, there is a wide range of  
6 interests, concerns, and differences among persons with disabilities. So that should be  
7 something that we should focus on as well.

8 DR. REEDE: Thank you.

9 Ms. Masny?

10 MS. MASNY: Yes, this is a comment, and then just a suggestion for one of  
11 the resolutions. In reviewing the papers from the responses from all the professional  
12 organizations, I do believe that almost every single one of the professional organizations did  
13 mention NCHPEG, and what the core competencies meant for them, and how that was one of  
14 the pieces that they were able to integrate into the work that they were doing to integrate  
15 genetics into practice, in genomics into practice.

16 One of the comments that we received from Dr. Jean Jenkins was to  
17 actually recognize NCHPEG and the work that they have done in part of the resolution. I think  
18 that it would fit very well into the second recommendation regarding the partnerships and the  
19 cataloging of information, and sharing information to actually mention NCHPEG there, both to  
20 hopefully get more visibility for NCHPEG, and maybe future members, as well as to see it as  
21 one of the models for partnership.

22 DR. REEDE: Thank you very much.

23 I want to thank all the members of the roundtable and the members of the  
24 task force for their work on this, and we'll use this in further deliberation. Thank you.

25 DR. McCABE: Thank you very much, Dr. Reede, and all of our invited  
26 guests for sharing with us the work of your organizations in advancing the education and  
27 training of health professionals in genetics and the insight on how to encourage more of these  
28 efforts.

29 Your responses, and all of the responses we've received to our information  
30 request and this roundtable discussion, are immensely helpful to our exploration of private  
31 sector efforts in genetics education and training, and certainly it will be very useful to us as we  
32 further develop our resolution to the Secretary on this important topic.

33 With that, we will be discussing the draft resolution during a working lunch,  
34 and so to all committee members and ex officio members, box lunches are available for you out  
35 in the hallway. Please take a few minutes -- literally a few minutes -- to gather your lunches.

36 For members of the public, lunch is available in the hotel restaurant, which  
37 is on the way to the lobby.

38 We will reconvene in about 10 minutes.

39 (Whereupon, at 12:15 p.m., the meeting was recessed for lunch, to  
40 reconvene at 12:25 p.m.)

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1 AFTERNOON SESSION (12:36 p.m.)

2 DR. McCABE: We're going to consider the draft resolution on genetics  
3 education and training of health professionals. This can be found at Tab 4 of your briefing  
4 book, and staff has also put it up on the screen here. Joan, are you ready? I didn't check with  
5 you. I'm going to turn it over to you for this discussion of the education resolution, or we can  
6 do a tag team so we can both eat, if you'd like to do that.

7 DR. REEDE: Why don't we tag while I chew?

8 DR. McCABE: Okay. So I'll start off, and then Joan will take over. You  
9 have the draft resolution up before you. Somebody commented that it sounded a bit stilted to  
10 have all the whereas's, but unfortunately, that is the nature of resolutions, to have all those  
11 whereas's, so I think we're stuck with those.

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

16 I know it has been brought up about family history being the first genetic  
17 test. I think that a couple of the ex officios were looking at where we might fit family history  
18 into this. Any comments? Alan, were you one of the ones working on that?

19 DR. GUTTMACHER: I wasn't working specifically on a place to put it, but  
20 I think there were a number of us that thought it should be put somewhere.

21 DR. McCABE: Okay.

22 DR. GUTTMACHER: But I will pay attention now while we work over  
23 lunch as to where it should be put, unless somebody else has a good idea.

24 DR. McCABE: We could certainly work on including it in the  
25 recommendations, if we didn't want to insert it in the whereas.

26 DR. GUTTMACHER: And I would think it would make more sense as a  
27 recommendation, rather than a whereas.

28 DR. McCABE: Robinsue?

29 DR. FROHBOESE: I do have a specific recommendation. I think in  
30 keeping with the very good recommendation that Reed first came up with, and then others  
31 supported in linking with the Secretary's efforts on health information technology, and the fact  
32 that that could be one place where we can include family information, I think in  
33 recommendation Number 1 which already addresses departmental policies and programs about  
34 genetic information, that there we specifically include family history, and a recommendation  
35 that the CAHIT effort in the  
36 Department on health information technology include looking at genetic information and family  
37 history as part of its overall recommendations in formulating an action plan.

38 DR. McCABE: Okay. Joan, are you ready to take over?

39 DR. REEDE: Sure. Moving forward, any other comments on incorporation  
40 of something related to family history and looking at inclusion of information technology,  
41 looking at genetics and family history within recommendation one?

42 DR. KHOURY: Maybe this was said earlier. But I guess anytime we use  
43 the word "genetic information," we can put slash, family history to it, because family history is  
44 much more than genetics. It represents shared environment, shared cultures, and shared  
45 behavior.

1                   So the philosophy that genetic information and family history should not be  
2 exceptional and so on and so forth, you know, search for the word genetic information in family  
3 history.

4                   DR. REEDE: Martin?

5                   MR. DANNENFELSER: I guess I just wondered, in the context of the  
6 Genetic Information Nondiscrimination Act, is this covered information within that, or not?  
7 Obviously if you have information in a medical record relating to preexisting conditions, that  
8 can be problematic, I imagine, to a person in terms of health insurance coverage, or potentially  
9 employment. Does putting family history in there have any impact on whether or not that is  
10 protected information within the context of the Genetic Information Nondiscrimination Act?

11                  DR. REEDE: Any comments?

12                  MR. GRAY: Well, the bill, as passed by the Senate, defines genetic  
13 information to include family medical history. It is specifically included in that, and I think it is  
14 important that we recognize family medical history is a part of genetic information. I would  
15 certainly make the point.

16                  DR. REEDE: Any other comments with regard to this family history and  
17 information technology in recommendation one?

18                  DR. FROHBOESE: I was just going to add that by including it in that first  
19 recommendation, and in the context of the Secretary's Health Information Technology effort, a  
20 critical part of what that group is doing is looking at privacy, confidentiality, and  
21 nondiscrimination. The Office for Civil Rights is an active participant in that, and is ensuring  
22 that the privacy rule under HIPAA and nondiscrimination considerations are part of the  
23 formulation.

24                  So I think by putting it in that first recommendation, we'll have some  
25 assurances that those considerations will be part of formulating any recommendations.

26                  MR. GRAY: Can I just make one suggestion? The way it is written now, it  
27 says, "genetic information, including family medical history information." I would just change  
28 that to say, "which includes family history information."

29                  The reason for that is that I want to make sure it is clear that family medical  
30 history is a part of genetic information, that is not something that is separate.

31                  DR. REEDE: Moving on, the next whereas is, "Advances in genomics have  
32 the potential to greatly improve health status and outcomes," and "Appropriate and adequate  
33 training and education in genomics is crucial for all health professionals to assure the  
34 successful integration of genomic concepts and genetic technologies, and services throughout  
35 the entire health care system."

36                  DR. LEONARD: Maybe we should add to that, "Whereas health  
37 professionals and the public." Because we have been bringing up over and over again that the  
38 public is an active part of this process, and so their education is just as important as the health  
39 professionals, in a different way.

40                  DR. REEDE: Any other comments?

41                  MS. HARRISON: Joan? I would advocate that we make that a separate  
42 whereas, because I think the adequate training and education in genomics, obviously we're  
43 meaning a different type of training and education. So I very much advocate for the public  
44 education piece to be in the whereas, but I think it should be separate.

45                  DR. REEDE: Any other specific comments with regard to the public

1 education piece? I think there was actually a very rich discussion at the end about the need for  
2 the public education. It seems clear that it should be incorporated here somewhere in terms of a  
3 separate whereas, or an integration into these current ones. Any thoughts?

4 DR. WINN-DEEN: I think it is important to include sort of the public  
5 education and capture this concept as an informed consumer, that we need them to be educated  
6 so that they can be informed consumers, and active participants in their health care.

7 DR. FEETHAM: For the previous one, to be consistent with the language  
8 from Muin's comments, and Toby Citrin's comments of saying health care and public health  
9 professionals in your prior whereas, and look at that throughout the document.

10 DR. REEDE: So in this area, and any other area?

11 DR. FEETHAM: Pardon?

12 DR. REEDE: So in this section, but in any other areas, making it more  
13 broad?

14 DR. FEETHAM: Right. Saying health care and public health  
15 professionals, I think you'll cover a broader scope.

16 DR. REEDE: The recommendation here was to, "Engage other health  
17 professionals, the private sector and colleagues at the federal and state levels to facilitate the  
18 cataloging and dissemination of genomics applications to clinical medicine and public health,  
19 and models based upon these applications to ensure genomics has impact now, as opposed to  
20 far-off in the future." Emily?

21 DR. WINN-DEEN: I just want to say I think before we move down to  
22 number two, that we should probably split one into two different things. One is sort of  
23 including genetic stuff and Department policies, and then the second part of that is talking  
24 about education. Then I think two becomes three then, if we do that. It just seems like one  
25 now has sort of two different thoughts in it.

26 DR. REEDE: So one relates to departmental policy, and the other one  
27 relates to overall education and training. Okay.

28 MS. CARR: And where would you see the point about family history?  
29 Would that be part of the second one, then? Or a separate one altogether?

30 DR. WINN-DEEN: I think the family history, it needs to be integrated as  
31 part of the training and education of health professionals to take family history, and to make it  
32 part of medical records. I think it is appropriate, sort of the way you have split it there.

33 MS. CARR: Okay.

34 DR. REEDE: Look at recommendation Number 2.

35 DR. LEONARD: And so two should start with "Integrate genomics into the  
36 education." So it is like the other ones.

37 DR. TUCKSON: (Inaudible.)

38 DR. McCABE: Reed, we're not hearing you. You'll need to speak up.

39 DR. TUCKSON: The third whereas. Can you hear me now?

40 DR. REEDE: Yes.

41 DR. McCABE: Reed? It sounds like you are a little bit close to the mike.

42 DR. TUCKSON: How about now?

43 DR. McCABE: That seems a little bit better.

44 DR. TUCKSON: (Inaudible.)

45 DR. McCABE: You're breaking up.



1 DR. TUCKSON: I'm breaking up now?  
2 DR. McCABE: And we aren't trying to ignore you by saying that.  
3 DR. TUCKSON: All right. I'll try to call on a different line.  
4 DR. McCABE: No, that's better.  
5 DR. TUCKSON: Okay. The third whereas --  
6 DR. REEDE: I beg your pardon?  
7 DR. TUCKSON: I heard you a moment ago about the third whereas  
8 (inaudible.)  
9 DR. REEDE: Right.  
10 DR. TUCKSON: Have you completed that discussion?  
11 DR. REEDE: No, I don't believe we have. We talked about the addition of  
12 the public, and putting it within this whereas, or creating a separate whereas that refers to the  
13 need for public education and training.  
14 DR. TUCKSON: I'd like to urge a little bit of (inaudible) integration, add a  
15 little bit of that to the discussion.  
16 DR. REEDE: No, it hasn't. So you're speaking specifically to the word  
17 "successful" and another word that might be more suitable?  
18 DR. TUCKSON: Yes, and I'm not sure what successful means. And so I'm  
19 thinking of appropriate, effective, and efficient integration.  
20 DR. REEDE: Appropriate is what we have up there now.  
21 DR. TUCKSON: Appropriate, effective, and efficient.  
22 DR. REEDE: Is there any further discussion around incorporation of the  
23 public within this, or setting up a separate whereas for public education?  
24 DR. FELIX-AARON: I think as a standalone whereas, I think it is very  
25 effective. The only other thing you may want to consider is to add a phrase at the end. It could  
26 say, "Whereas appropriate education and genomics is crucial for the general public to reap the  
27 benefits of, or to take advantage of the power of this." So that it clearly specifies that not only  
28 the public needs to be engaged to be better consumers, but also for them to get the benefits of  
29 this vast technology.  
30 DR. REEDE: Okay. The next whereas was "Whereas such integration is a  
31 necessary component of access."  
32 MS. CARR: Dr. Reede, can I just go back to the one prior to the general  
33 public? The last part of that read, "the entire health care system," and we had a suggestion that  
34 it just read "the entire health system," that that might broaden it a bit. Is that okay?  
35 DR. LEONARD: In the second whereas, I guess it is, the third whereas, we  
36 were going to add public health and health professionals, or we were going to broaden that to  
37 be public health and health professionals.  
38 DR. FEETHAM: The language that we used in a lot of our documents  
39 through HRSA and NIH is health care and public health professionals.  
40 DR. REEDE: Brad?  
41 MR. MARGUS: I have another whereas to talk about. Are we ready for  
42 another whereas, or do we have to finish that whereas? Going to my question about what was  
43 broken, and Mr. Citrin's response to it, I was thinking that this might sound a little negative, but  
44 it would still underscore maybe an urgency to do something.  
45 Something along the lines of, "Whereas insufficient training and education

1 can lead to excessive costs incurred on inappropriate tests," since he brought that up,  
2 "inaccurate, or at least illusive disease diagnoses," which is what we just talked about, "and in  
3 many cases, misguided disease management and family planning." Those are all negative  
4 things that can come about if the Secretary doesn't do anything about this.

5 DR. REEDE: Sort of in this area, the broken things, the issues that created  
6 urgency. Are there others that you would want to add to that list?

7 DR. WINN-DEEN: Yes. When Sarah gets done typing this list, and I'm  
8 just going to comment that I don't think the excessive costs should be the first thing. I think the  
9 first thing should be the patient management issue. Obviously costs are important, but I think  
10 the key thing is that patients are not being managed as they should be for their medical  
11 condition.

12 DR. FEETHAM: Another concept of that lack of education is access, in  
13 addition to the patient management.

14 MS. BERRY: We'll probably have to reorder them, won't we? Because  
15 when it says, "such integration," it refers to the two other whereas' above.

16 DR. REEDE: What you said is education is a necessary component of  
17 access. I was wondering if you wanted to say something more specific about genetics.

18 MS. ZELLMER: On the whereas as far as where it says, "Such integration  
19 is a necessary component of access," which should refer to the third one, I think actually access  
20 is affected by education of not only the health care professional, but the public. So maybe we  
21 could just say, where education is a necessary component of access, rather than integration.

22 DR. REEDE: Or possibly mention both the public and the health  
23 professionals, sort of public health mentions specifically the education of this body as an  
24 integral component.

25 MS. CARR: Tell me what to do. I was so engrossed in that.

26 MS. ZELLMER: Under the next whereas, instead of "such integration," say  
27 "education of health care and public health professionals, and of the public is a necessary  
28 component of access." Does that sound right?

29 DR. REEDE: I'm sorry. Either one.

30 DR. FELIX-AARON: The fourth whereas, or Brad's whereas. I think one  
31 of the points that Mr. Citrin made that wasn't reflected in this whereas is you name cost, sort of  
32 the media distortion, and the role of expanding technology that can either reduce disparities, or  
33 exacerbate disparities. I'm not sure that whereas, that third point, was captured in that whereas.

34 DR. REEDE: You want to offer some wording?

35 DR. FELIX-AARON: Exacerbate disparities, health disparities.

36 MS. CARR: This is a --

37 DR. FELIX-AARON: I'm not asking for there to be a separate whereas.

38 DR. REEDE: It is just another item to be mentioned.

39 DR. FELIX-AARON: Sort of the urgent need for this.

40 MR. MARGUS: It sort of belongs maybe in the next whereas, related to  
41 access to.

42 DR. FELIX-AARON: Where is education?

43 MR. MARGUS: Isn't there a place where we talk about new technologies?

44 DR. REEDE: No, actually, I think the issue around exacerbation of health  
45 disparities belongs up with the other urgent areas.

1 DR. FELIX-AARON: Yes. Sort of where we talk about the impending  
2 crashes.

3 DR. REEDE: Down? Where insufficient and training can lead to?  
4 DR. FELIX-AARON: Right. So whereas insufficient education and  
5 training can lead to inaccurate disease diagnosis, and misguided management.

6 MS. CARR: Do you want it before costs, or after?  
7 DR. REEDE: Before cost. Last, okay.  
8 Muin?

9 DR. KHOURY: The idea captured here is evidence-based health care  
10 prevention. When we talk about access and education of public health professionals  
11 (inaudible), we're saying education is very important for the evidence-based practice of  
12 medicine. So maybe we can repackage that paragraph in a positive light or a negative light,  
13 because we're talking about sufficient education can lead to that stuff. (Inaudible.)

14 DR. REEDE: That was going to be my question. How would you massage  
15 this?

16 DR. KHOURY: I mean, just the same thing, a necessary component of  
17 access, a necessary component of evidence-based practice of medicine and health care,  
18 whatever you want to say. Really, education is a component of for various reasons, not only  
19 access.

20 MS. CARR: Muin, are you saying that should replace the "Whereas,  
21 insufficient education"?

22 DR. KHOURY: Yes, or put it somewhere in there.

23 DR. WINN-DEEN: Sarah, why don't you just put education and training  
24 prevent inaccurate or (inaudible) disease, so it is not a negative.

25 DR. KHOURY: If I could use the word  
26 "evidence-based," that's all. If we can find a place for it, that would be nice.

27 MS. CARR: Well, and I guess positives are always better than negatives,  
28 but I think Brad was sort of -- well, it is in response to your own concern that we need to call  
29 attention to a problem, I guess.

30 DR. WINN-DEEN: Well, what about if we just add the evidence-based  
31 medicine thing in the one that you were working on that ends in "of access," evidence-based  
32 medicine improved prevention and public health, and whatever buzz words we want to put in  
33 there.

34 MS. MASNY: Or we could put it in the higher up with the, "Whereas  
35 advances in genomics have the potential to move us to a preventive model of health and  
36 evidence-based practice to greatly improve the health status." Because that is going to be the  
37 mechanism by which we're going to improve the health status and outcomes.

38 DR. HACKETT: If you're talking about  
39 evidence-based medicine, isn't that entirely a third new topic that might confuse things? We're  
40 talking about genomics, genetics, and then evidence-based medicine.

41 MS. MASNY: I would think that the implication there would be that now  
42 that we understand the biology of the disease, or conditions, that that is going to be the  
43 evidence base. So I think it goes hand in hand with the genetics and genomics.

44 DR. HACKETT: But that is such a big topic by itself, is the only concern.  
45 DR. REEDE: I think it is written where it is the application of evidence-

1 based medicine. I have no problem with it being printed that way. Do others want to weigh in?

2 MS. CARR: What about what Agnes was suggesting? Were you saying  
3 that we should modify the second whereas in some way? Or does this take care of it?

4 MS. MASNY: That is what I was suggesting for the second one, because I  
5 think that the issue of the preventive model that was suggested, as well as  
6 evidence-based is the actual cause, or the thing that precedes the health status and outcomes,  
7 much more so than related to access.

8 MS. CARR: What would you suggest, then?

9 MS. MASNY: Just that whereas advances in genomics have the potential to  
10 provide evidence-based practice, and greatly improve health status and outcomes.

11 DR. REEDE: Any other comments on the whereas' so far?

12 DR. WINN-DEEN: Do we want to say  
13 evidence-based practice in medicine, or something, instead of just evidence-based practice?  
14 Practice of what?

15 DR. TUCKSON: (Inaudible.)

16 DR. REEDE: Reed, we can't hear you.

17 DR. TUCKSON: Can you hear me?

18 DR. REEDE: Yes.

19 DR. TUCKSON: (Inaudible.) How does that read now?

20 DR. REEDE: How does it read now? I'll read it for you. "Whereas

21 advances in genomics have the potential to facilitate evidence-based practice of medicine and  
22 greatly improve health status and outcomes."

23 DR. TUCKSON: The difficulty here is needing the word evidence-based  
24 practice. The genetics doesn't facilitate evidence-based practice. What is the evidence for the  
25 (inaudible) knowledge of whatever the discipline is. So advanced genetics (inaudible) into the  
26 guidelines and the actual practice of (inaudible).

27 DR. KHOURY: I agree with Reed. I agree with what Reed Tuckson just  
28 said.

29 DR. REEDE: Reed, would you like to make a suggestion for how we might  
30 rephrase this?

31 DR. TUCKSON: I think if I understand (inaudible) advances in genetics  
32 will lead to more (inaudible) understanding of disease process issues, and will (inaudible)  
33 better guidelines around what interventions genetic (inaudible) effective, and appropriate.

34 MS. CARR: Reed, we have modified it along the lines you suggested. So  
35 now it reads, and just make sure it is what you suggested. "Whereas advances in genomics will  
36 lead to more precise understanding of disease processes, and will provide better guidance on  
37 the application of therapeutic and preventive strategies," and I guess I lost you after that.

38 DR. TUCKSON: No, that's much better.

39 MS. CARR: Okay.

40 DR. LEONARD: But then can it just read, "to greatly improve health status  
41 and outcomes?" So take out, "have the potential to facilitate," right?

42 DR. REEDE: We have incorporated evidence-based practice in another  
43 section. Do you want to read that one again, also?

44 MS. CARR: Okay.

45 DR. McCABE: Before we leave that one, the editor in me sees a split

1 infinitive. Can we take the "greatly" out, please?

2 MS. CARR: Okay. The second reference to evidence-based medicine  
3 reads, "Whereas education of health care and public health professionals and the public is a  
4 necessary component of access and the application of evidence-based medicine."

5 DR. McCABE: Reed, could you mute the phone in between, please? We're  
6 getting some feedback and echo.

7 DR. LEONARD: That's like two concepts in one whereas. I don't see what  
8 access has to do with the application of evidence-based medicine. Those are two different  
9 concepts, and they need to either be split or --

10 DR. REEDE: Dr. Khoury?

11 DR. KHOURY: Why don't you split them? Have two whereas'.

12 DR. REEDE: Is there agreement on this? Okay. The next whereas reads  
13 as, "Whereas, through a survey of Federal agencies on their role and activities in genetics  
14 education, training, and health workforce analysis, it was found that Federal efforts are focused  
15 on enhancing access through facilitating the translation and appropriate integration of new  
16 genetic technologies into health care and public health."

17 DR. KHOURY: Is that all the federal survey found was related only to  
18 enhancing access? Or everything else? It seems to me that there was more than that, and  
19 maybe I missed that in the last meeting when you guys discussed this. But it seems like the  
20 focus of the federal efforts is not only on enhancing access, but a whole bunch of other things  
21 as well.

22 MS. CARR: Well, I think this is probably a distillation and sort of boiling it  
23 all down. But if in doing this, we have neglected to highlight something important, we can just  
24 add it here, I think, if the committee agrees.

25 DR. KHOURY: Well, the whole concept of translation and appropriate  
26 integration includes access. So if you just say that the federal efforts are focused on translation  
27 and appropriate integration, because without just highlighting enhancing access, it seems to me  
28 that it is too focused. Just take away "enhanced access." That's it.

29 DR. McCABE: Do the other federal agencies agree with that? Anyone  
30 who disagrees?

31 MS. CARR: Well, the access agency isn't in the room at the moment, so we  
32 might want to come back to that.

33 DR. HACKETT: A quick question. Would you want to change that to  
34 genomics, or just leave it as genetics? Both the first and last sentences.

35 DR. REEDE: Genetics and genomics?

36 DR. McCABE: I would make it genetics and genomics, I think.

37 DR. REEDE: I would suggest that when HRSA returns, we revisit this.

38 The next whereas is, "A solicitation of information from educational and professional  
39 organizations, identified the following needs in genetics/genomics education and training."  
40 Those needs were identified as, "Inventoried, widely relevant clinical applications stemming  
41 from advances in genomics; educational models that use such applications to clarify how  
42 genetics/genomics should be integrated into practice; a broadening of the focus of education  
43 and training from genetics to genomics; appropriately trained faculty; and training programs  
44 that address the interface of, and interaction between, genomics and public policy." Khoury?

45 DR. KHOURY: I would suggest to revisit family history here as under its

1 own bullet, or one of these bullets. But it could be its own bullet.

2 MS. CARR: Well, I think we'd have to make sure that the information that  
3 came back from these organizations, that is what this bullet is about. Amanda or Joan, was the  
4 family history highlighted in the survey response?

5 DR. REEDE: No, it wasn't mentioned there. But one of my questions  
6 would be, this is a solicitation of information. If you consider it a solicitation that included the  
7 survey and the roundtable, in that case, it could be included.

8 MS. CARR: And the two changes that are here are reflective of comments  
9 made by the roundtable, too. So how would you like to add it, and where?

10 DR. KHOURY: It could be under the third bullet, "A broadening of the  
11 focus of education and training from genetics to genomics, including."

12 MS. CARR: Which includes the point Peter --

13 DR. KHOURY: Which includes family history tools.

14 DR. REEDE: One of the other areas that was mentioned, I think, by Dr.  
15 Whitcomb, in addition to the clinical applications, he talked about the need to access patients,  
16 and this isn't reflected here. Do we want it to reflect that?

17 MS. BERRY: I don't know if it's accurate to say, but this gets to the point  
18 about how significant is the problem. Is it accurate to put in the beginning part before the  
19 bullets that these are urgent needs? Use that modifier? I don't know if everyone would agree  
20 that every one of those is urgent, but that would sort of add some oomph to the resolution, if  
21 you think that is accurate.

22 DR. LEONARD: Can I make a suggestion in the bullets? The family  
23 history tool is really more a part of the second bullet, which is integrating genetics and  
24 genomics into practice. The way that you're going to do that in general practice is by use of  
25 family history tools, more than a broadening of the focus of education and training from  
26 genetics to genomics.

27 MS. CARR: And maybe in this case, it could read "including," rather than  
28 "which includes."

29 DR. LEONARD: Or "through the use of." Well, yeah, you could just say  
30 "including," or "through the use of." Whatever.

31 DR. REEDE: One of the areas that was mentioned in the comments related  
32 to the use of the Internet technology and those types of things, and I'm wondering if there is not  
33 a way to integrate some of that, along with the information technology that we're talking about  
34 here. The importance of this is sort of the future area.

35 DR. LEONARD: Well, it could be through the use of family history tools,  
36 information technologies, and  
37 web-based practice tools, or things like that. That is basically what we were talking about, as a  
38 way to get it into the hands of practicing physicians, which I think is what this second bullet is  
39 really getting at.

40 DR. REEDE: I think it's fine. I think the more we can link the concepts  
41 across, and since information technology came out so much today, and it was mentioned in  
42 another way, but it was mentioned in the responses, to have it incorporated here would be  
43 useful.

44 Agnes?

45 MS. MASNY: Under these bulleted items, during the roundtables, it was

1 mentioned several times about the importance of the certification, licensure requirements, and  
2 things like that. Someone had mentioned that it should be included in the whereas as to why  
3 education should be mandated.

4 So since the certification question was one that we had with all the survey  
5 participants, as well as in the roundtable, if the group thinks that a separate bullet item about  
6 the importance of mandating --

7 DR. LEONARD: Well, I think the point was made that unless you mandate  
8 it in the certification or licensure, whatever, by those bodies, it is not going to change in the  
9 curriculum.

10 DR. REEDE: I'm wondering if a way, since so much of that is occurring at  
11 the state level, or through private organizations and not really the government level, for part of  
12 the certification licensing, so the licensing is occurring at the state level, I'm wondering if  
13 there --

14 DR. LEONARD: But accreditation is more at a national level, so there are  
15 accreditation bodies. The licensing is more state by state.

16 DR. REEDE: Right. So I'm wondering, just being very specific, and not  
17 sort of putting the licensing, credentialing, and accreditation all together, because it is difficult  
18 to mandate that from a level that does not really control that area. So are you speaking  
19 specifically to the accreditation here? A bullet specific to accreditation?

20 DR. LEONARD: Well, I actually found it very frustrating to know that for  
21 physicians, that there is nothing that can be done nationally. That is pretty pathetic, frankly.

22 DR. McCABE: Well, I think what can be done nationally is to work with  
23 our boards, our professional boards, and those are the organizations that set standards  
24 nationally.

25 DR. LEONARD: And that's more accreditation, rather than licensure. So  
26 maybe if we want to put something in about national accreditation standards that, or the need  
27 for national accreditation standards to drive the incorporation of genetics into the curricula of  
28 educational bodies, or whatever. I don't know. I can't say that again, though.

29 DR. McCABE: Then make sure we put accrediting and reaccrediting,  
30 because in fact it is the reaccrediting that is required these days that is even more powerful in  
31 keeping the standards up.

32 DR. REEDE: The reaccrediting also addresses parts of the issues that you  
33 raised about what do you do about physicians and health providers that are already in practice.

34 MS. CARR: I'm sorry, but I didn't get that.

35 DR. LEONARD: It would be need for --

36 MS. CARR: I'll just say "need" at the end, but we have national --  
37 remember, this is the urgent needs of the --

38 DR. LEONARD: Okay. National accreditation standards. Accreditation  
39 and reaccreditation standards, sorry.

40 PARTICIPANT: "Accreditation" is a facility term. I think you're meaning  
41 "certification."

42 DR. McCABE: That's right. It's certification, and then recertification.

43 MS. CARR: So are we changing this?

44 DR. LEONARD: Yes. "Certification and recertification." "Standards for  
45 genetic education, or genetic and genomic education to drive" -- or is it education or

1 proficiencies? Or competencies? "Genetic and genomic competencies to drive the" -- I don't  
2 know what you want to call drive.

3 MS. CARR: Influence?

4 DR. LEONARD: "Influence the incorporation of genetics and genomics in  
5 educational curricula."

6 DR. REEDE: I have a question. Is this better under a whereas, or should  
7 this be one of the recommendations that should be put forth?

8 DR. LEONARD: Well, this is something that they identified, at least in the  
9 discussion here, at least one person said that when there are mandates to provide certain types  
10 of education, that is what drives the curriculum changes.

11 So we may want to sort of take that as something that we could resolve to  
12 do also, but that was something that they were saying is a need. If you're going to change the  
13 curricula for many health professionals, then it helps to have accreditation standards that  
14 include genetics.

15 DR. WINN-DEEN: I think the other point was that we're not looking for  
16 people to become certified in genetics. We are looking for them to have standards for genetic  
17 and genomic competencies in order to influence the incorporation of genetics into genomics in  
18 educational criteria across disease areas, or across practice areas, sort of that concept that we're  
19 not just looking for some specialty in genetics, but that this crosses all medical disciplines.

20 DR. REEDE: So across disciplines?

21 DR. WINN-DEEN: Yes.

22 DR. LEONARD: Or could you say genetics and genomics in general,  
23 health care education curricula, or something?

24 DR. WINN-DEEN: Yes, just so that we're clear that --

25 DR. LEONARD: You're not creating specialists. We're creating competent  
26 generalists.

27 DR. WINN-DEEN: Right, or whatever, competent subspecialists.

28 DR. REEDE: Any other whereas? Robinsue?

29 DR. FROHBOESE: Yes, I have another suggestion for an addition. I think  
30 another central theme that came out of the survey of organizations, and that we've heard this  
31 morning which is also reflected in the recommendations, is the cultural diversity within, and the  
32 cultural competency of health professionals in genetics or genomics. So I would suggest  
33 adding that as a bullet.

34 DR. LEONARD: But that is something that genetics health professionals  
35 do. Do you hear that being stated as something that needed to be done in general for all health  
36 care providers?

37 DR. REEDE: I think it was felt by all. It wasn't just for the specifics of  
38 genetics, it was across the board.

39 MS. CARR: Would that be incorporated into the appropriately trained  
40 faculty? Or that would apply to all practitioners as well?

41 DR. REEDE: All practitioners.

42 DR. McCABE: While Sarah is working on that, let me just do a little bit of  
43 housekeeping. That is that we're running over the allotted time for this, but I think that it is  
44 quite appropriate that we do that. The coverage and reimbursement, we probably are not going  
45 to complete discussion of this afternoon, and that will continue, I'm sure, into the next meeting.



1                   So I'm going to take the Chair's prerogative to extend this session. I just  
2 want to check, Reed, are you able to be with us for awhile?  
3                   DR. TUCKSON: (Inaudible.)  
4                   DR. McCABE: It sounds like you are commenting from outer space  
5 somewhere, Reed.  
6                   DR. TUCKSON: (Inaudible.)  
7                   DR. McCABE: Sorry. We really can't hear you.  
8                   DR. TUCKSON: Can you hear me now?  
9                   DR. McCABE: Not really. Is there any chance you can get to a land line,  
10 Reed?  
11                   DR. TUCKSON: (Inaudible.)  
12                   DR. McCABE: You sound really cool, but we can't understand a word of it.  
13 (Laughter.)  
14                   DR. TUCKSON: I'll call back on a land line.  
15                   DR. McCABE: Thank you.  
16                   DR. REEDE: Since HRSA is back in the room, there was one of the  
17 whereas' where we changed the wording and removed "enhancing access," and wanted you to  
18 weigh in on that, to read it, and give any comments.  
19                   DR. FEETHAM: I was here for that part of the discussion, and I think that  
20 clarifies and strengthens it.  
21                   DR. REEDE: Okay. Is that it for the whereas'? Can we move to the --  
22                   MS. HARRISON: I have one comment. If we can just go back to Brad's  
23 whereas. I wanted to try to make it even a little bit stronger. We had put "training can lead to,"  
24 and I was wondering if you could change it to "is leading," because we know that it is  
25 happening now from the two stories that people have said.  
26                   MR. MARGUS: It could be "has led."  
27                   MS. HARRISON: "Has led," or something like that. You know it is  
28 happening.  
29                   DR. REEDE: If we're in agreement, do you want to go through these again  
30 just briefly?  
31                   MS. HARRISON: I see you're scanning through the rest of them. I don't  
32 know if we can change other verbs in there.  
33                   DR. McCABE: We're probably going to have to have this edited. What we  
34 have done typically in the past, because it is hard to get all the grammar right while we're doing  
35 this. So Sarah, can I volunteer you and your staff to work on this this evening?  
36                   MS. CARR: Yes.  
37                   DR. McCABE: Okay. And then bring it back tomorrow. I think that will  
38 be a more efficient way to go to get as detailed as we can, but not to worry too much about the  
39 grammar, having tried to unsplit an infinitive myself.  
40                   DR. REEDE: It's more are there any other concepts that we should include  
41 within this section?  
42                   DR. McCABE: What I would suggest is that we go to the resolutions per  
43 se, and then do a quick readover at the end to see if there is anything we pick up, because the  
44 two may inform each other.  
45                   DR. REEDE: Resolutions. "As such, and in light of the importance of

1 ensuring that the benefits of the genetics/genomics revolution are accessible to all Americans,  
2 the committee urges the Secretary to take the following steps to ensure that genetics education  
3 and training of all health professionals is adequate." I think it would be, "All health and public  
4 health professionals," that would be changed.

5 The first as currently written. Let me turn, because these have been  
6 changing. Actually, Sarah, it is easier for you to read than for me to try to do this.

7 MS. CARR: Okay. Let me read the first one. "Promote and actively  
8 incorporate into departmental policies and programs the philosophy that genetic information,  
9 which includes family history information, should not be treated as exceptional, but rather as  
10 part of the spectrum of all health information."

11 DR. REEDE: One of the comments that was returned with regard to this is  
12 the question of talking about exceptionalism, and this language, to the extent to which it is  
13 compatible with the language that we're using when we talked about direct-to-consumer  
14 marketing and wanted to know if there was any comments.

15 MS. CARR: I think what you are saying is someone pointed out the fact  
16 that what we are suggesting in the direct-to-consumer marketing resolution is not compatible  
17 with this recommendation. I think in Reed Tuckson's comments on the coverage and  
18 reimbursement report, he also took note of a couple of places where it appears to recommend a  
19 more exceptionalist approach.

20 I think the committee in March, when addressing the issue of genetic  
21 exceptionalism, concluded that it might be necessary to assess the need for exceptionalism, or  
22 the problems associated with it on a case by case basis. So I think you have left yourselves  
23 room to be inconsistent, I guess.

24 DR. WINN-DEEN: I think in this case, what we're talking about is really  
25 sort of not so much that genetics is exceptional but that it happens to be the new thing that we  
26 have to integrate into medical practice today, and so it is just the latest thing which we need to  
27 deal with.

28 DR. LEONARD: I think also you're talking about genetics as a  
29 subspecialty, and moving it from subspecialty practice to influencing every health care and  
30 public health professional. So I think it is more that it is not a subspecialty, so we may not even  
31 want to use the word "exceptionalism," and talk about it as specialized as opposed to general  
32 practice. Genetics is going to influence all of medical practice, so it has got to be part of every  
33 health care professional's training.

34 DR. WINN-DEEN: So this should not be treated as exceptional, it should  
35 not be treated as a subspecialty, or as a specialty.

36 MS. CARR: Or what if you just said, "should be treated as part of the  
37 spectrum of all health information?"

38 DR. WINN-DEEN: Yes, that's fine. Just take the "not" and the  
39 "exceptional" out.

40 DR. LEONARD: Sarah, in the preamble paragraph, in the last sentence,  
41 can you change "all health professionals," to "all health care and public health professionals?"

42 DR. REEDE: Reed, are you back again?

43 (No response.)

44 MS. CARR: And the second one isn't really integrated yet completely, but  
45 this part was part of the prior one. "Promote the integration of genomics into the education and

1 training of all health professionals," and this is a fragment, I think, from the first one, "so it  
2 should be treated and viewed." Actually, I think it belongs up there.

3 So the second one is, "promote the integration of genomics into the  
4 education and training of all health professionals." Here is where we wanted to build in the  
5 suggestion that the Secretary incorporate in the health information technology initiative, some  
6 reference to family history, or incorporate some electronic family history tools into that  
7 initiative.

8 DR. McCABE: Sarah, I would suggest that we separate out "add" into  
9 another resolution. It seems like we're mixing education and the Secretary's initiative, plus it  
10 will make it more obvious to the Secretary if it is not buried as a second part of the second  
11 resolution.

12 DR. REEDE: Wherever we have "health professionals," if we could sort of  
13 in the wordsmithing later, change it to "health care" and "public health professionals."

14 DR. McCABE: Reed, are you back with us yet?

15 (No response.)

16 DR. HANS: Just on this one, I guess I have some concerns about, because  
17 the discussion about CAHIT was broader than what actually the topic of this resolution was of  
18 education and training for health professionals. It is a much broader topic than just that.

19 So I would suggest one of two things. One, to narrow this down to just talk  
20 about the educational components to be considered in CAHIT for genetics information. And  
21 then put the question to the committee, if you want to take up the broader issues of CAHIT in  
22 the interface of this committee, sort of as a separate something, whether it is a letter, or some  
23 other separate topic.

24 I'd just be concerned if you'd try and capture everything in this resolution,  
25 because it is a more narrow topic than this opportunity offers. So this, if you want to narrow it  
26 down, could say something, "incorporate family history, tools, and practitioner educational  
27 tools, support." Instead of "tools," use "support," Sarah. And then I'm not quite sure where  
28 we're going with privacy and confidentiality, but there are a whole bunch of issues that may not  
29 fit with education and training.

30 DR. LEONARD: Can I suggest that practitioner educational support, really  
31 what you're looking at is point of care educational support? Because practitioner may be  
32 misconstrued as CME, or continuing education, which you really want it at the point at which  
33 you're seeing the patient.

34 DR. REEDE: I think I would also change the order. So I'd have as what  
35 you have as Number 2, I'd move down.

36 MS. CARR: So it would be Number 4?

37 DR. REEDE: Well, it would become 3, and 3 would become 2. While  
38 you're doing that, I'm going to read the next recommendation. "Engage other health  
39 professionals, private sector, colleagues at the Federal and State level to facilitate the  
40 cataloging and dissemination of genomics applications to clinical medicine and public health,  
41 and models based upon these applications to ensure genomics has impact now, as opposed to  
42 far off in the future." Any comments on this?

43 DR. LEONARD: There was a suggestion during this discussion to  
44 recognize the efforts of NCHPEG in this point, and I don't know how we do that.

45 DR. WINN-DEEN: Well, we could just say engage health professionals,

1 such as NCHPEG. Use it as an example.

2 MS. MASNY: But I think here, it is even stronger than just to engage them.

3 I think some of the resources we heard about, both through HRSA and through NCHPEG, that  
4 what we want to make sure is that these resources are utilized, and that the public, and even the  
5 professional organizations and academic institutions know about them, so they don't have to  
6 reinvent the wheel. So somehow if we could make it stronger in terms of utilizing existing  
7 resources that have been developed by state and federal organizations.

8 DR. REEDE: Giving NCHPEG as an example?

9 MS. MASNY: Yes. We could give NCHPEG and HRSA as an example.

10 DR. McCABE: I would just argue private, state, and federal organizations.

11 DR. REEDE: And not use an example?

12 DR. McCABE: It's fine to use an example, I just think we need to recognize  
13 the private sector.

14 DR. REEDE: Kay?

15 DR. FELIX-AARON: In reading this point, I agree with it in principle and  
16 what it says, but I was just struggling with sort of what the federal rule is here in terms of if we  
17 have an audience, and I imagine this is directed to the Secretary, how do we want to use that  
18 space that we have with the Secretary?

19 So I would think like for matters of just being effective as a document, as  
20 well as being efficient, I think this needs to be done, but sort of what the Secretary's role here  
21 is, isn't readily apparent to me. So I'd just like somebody around the committee to just clarify  
22 for me what the Secretary's role is here.

23 DR. WINN-DEEN: Well, it seems like it is sort of covered in the next point  
24 after this one, where we talk about specifically federal.

25 DR. FELIX-AARON: Right. I mean, that is one of the rules that the  
26 federal government has provided technical assistance, and I think the federal role is to do things  
27 that cannot be ordinarily done in the private sector because there is not the will, or there is not  
28 adequate incentive to do it. It is not clear to me that that recommendation captures the unique  
29 federal role.

30 DR. WINN-DEEN: So maybe what we want to say is partner with the  
31 private sector and state organizations, and encourage --

32 DR. FELIX-AARON: I think the one below says to support efforts. The  
33 point below, provide adequate program and technical support, but it is not clear to me why the  
34 federal government should be engaged in inventory and the cataloging process.

35 DR. LEONARD: Well, actually, the CDC is doing that, aren't you, Muin?  
36 You're cataloging the most common genes, and the most common diseases, and so in a sense,  
37 you are providing some of that information that would take genetics and genomics to the family  
38 practitioner.

39 DR. KHOURY: I think what I'm hearing Kay say here is that you need to  
40 give the Secretary sort of the best possible chance for integrating genomics, genetics, and  
41 family history into whatever Department-wide initiatives are going on. I think under Number 2,  
42 the Secretary's Health Information Technology Initiative is something that he can take and say  
43 okay, I'm going to integrate family history tools into that. But there are a number of other  
44 initiatives that the Department has. For example, the STEPS initiative, which is a big initiative  
45 for the prevention of common chronic diseases, and health promotion. That is really prevention

1 oriented.

2 So by encouraging the Secretary to look across the board and say okay,  
3 these are the initiatives that I own, that I've started, but so far I haven't seen the role of genetics  
4 and family history in it. But now, there are more legs I could stand on and use these as forums  
5 to empower big initiatives that use and integrate genetics, rather than be a standalone activity  
6 for the Department.

7 And so I think singling out the Health Information Technology Initiative is  
8 one thing, but I don't think we should focus on just one activity from coming out from HHS, we  
9 should give them a broad range, a number of activities that could touch the lives of all the  
10 agencies by using the concept of integrating family history tools and genetics into whatever we  
11 do, because that will affect the practice of health care, disease prevention, and health  
12 promotion.

13 That would include that last point, which is what Debra was talking about,  
14 which was what was the point about the cataloging function? It is only a minor tool, or a minor  
15 service that the Department can provide, but I think you should give the Department the  
16 broadest possible advice that affects most of the agencies, rather than one or two. That would  
17 be my advice.

18 DR. FELIX-AARON: I mean, I agree with that, but I also think that the  
19 point that I was trying to make in terms of to me, this seems like a small, narrow, small,  
20 function, and not necessarily something that the Secretary in his unique role would necessarily  
21 engage in. So CDC is doing it out of the programs, and I understand that.

22 But I agree with you in terms of providing vehicles. So to the extent  
23 possible, to provide vehicles where those types of recommendations can be tapped on, I think is  
24 extremely important.

25 DR. REEDE: Can I ask a question there? So is there a role sort of  
26 somewhere in between these, as we look at these multiple federal efforts? And having some  
27 place or some mechanism for understanding what is being learned within those efforts, and to  
28 link that back to the private sector, and to the educational sectors? So that as this work is being  
29 done, it doesn't end up being done in isolation, but the federal government is actually cataloging  
30 the work that it is doing in a way that it can be disseminated easily across these groups.  
31 Slightly different, but I think if you're not sitting in this room, you may not know about the  
32 work that is being done across the multiple agencies.

33 DR. FELIX-AARON: I mean, one of the things that come out of this  
34 Department and the Secretary is the issue of coordination, and coordination around the  
35 departments, and speaking with one voice. So I think, if what we have here could be framed in  
36 terms of speaking with one voice, or one Department and really understanding to make our  
37 Department more efficient and effective in that area, that resonates, at least in my mind, with  
38 what I hear coming out of the Department.

39 DR. McCABE: Well, if it read, "To coordinate and disseminate genomics  
40 information," would that be better?

41 DR. FELIX-AARON: I think so. I mean, I don't know what other people  
42 around the table think, but I would that at least at a minimum, it would. It sort of captures some  
43 of the conversations that have been going on here, and it resonates with what I hear coming out  
44 of the Department, and at least the goals of the Department.

45 DR. TURNER: I was just sort of agreeing that to determine what the

1 Secretary's role is is important, so that we can phrase these in ways that he is able to respond.  
2 In addition to what she is saying, I'm struggling with the softness of the language. Someone  
3 earlier suggested that we stand up a little taller and say, we may not want to go as far as direct,  
4 the integration of genomics into the education, because I don't think he can direct things.

5 But to look at where we are, and then sort of envision how would he carry  
6 this out, I think the words she just mentioned are the operative words, if you will, that would  
7 get the job done. So I would suggest that we think in terms of, do we want him to make a  
8 phone call? Or do we want him to take this to some meeting? Or how do we want this to play  
9 out?

10 DR. REEDE: So coordination, dissemination, and pulling it together.  
11 Debra?

12 DR. LEONARD: From this discussion, can we kind of take Number 2 up  
13 there, and I think what I'm hearing is that we pulled out one initiative, but there are steps, and  
14 there are other things, other initiatives. So maybe we should say incorporate genetics, family  
15 history tools, point of care, and educational support in the initiatives of the Department of  
16 Health and Human Services. For example, CAHIT, STEPS, that type of thing, and coordinate  
17 the dissemination of this into practice through private, state, and federal efforts.

18 But get in there that you want these things developed through the  
19 Department of Health and Human Services, and then you want that information that's  
20 developed there disseminated into practice. That is going to involve coordination and use of  
21 this information by professional organizations.

22 DR. REEDE: All right. I hear what you're saying. One of my questions for  
23 the agencies is the extent to which they can coordinate how the professional societies actually  
24 use this, as opposed to a coordination of how they disseminate the information that they have  
25 gathered. So I sort of see sort of a collecting what is going on, and being able to disseminate  
26 that, and to work with other organizations, but not really being able to coordinate after that, the  
27 activities of those professional organizations.

28 DR. FEETHAM: I think moving the discussion that we're having now  
29 about showing and giving the Secretary the guidance to look at this within all those key  
30 initiatives is really critical, and is a broader scope than adding it into some of these more  
31 specific processes. You're closer to that in Number 1, and promote and actively incorporate,  
32 and I think if you bring this overall concept of how this fits within many, or probably all of the  
33 Secretary's initiatives, I see as higher up the list, and more of an overriding. From then on,  
34 you're showing him some examples.

35 But I think you are losing it if you try to add all of those things, such as,  
36 such as, but I would bring it up, and I think it is closer to your current number one of  
37 incorporating interdepartment policies, but you're really talking about Department initiatives.

38 DR. REEDE: Ed?

39 DR. McCABE: I would ask that we try and get the big points down over  
40 the next 10 minutes, and then wrap this up. We're going to then take like a three to five minute  
41 break to try and make sure that we can get back to Reed and have him hear us, as well as us  
42 hearing him, and then proceed on. But if we can get the big ideas, we'll try and rough them out  
43 this evening.

44 DR. REEDE: Okay.

45 DR. LEONARD: Another big idea that needed to be added as a separate

1 bullet point was the education of the public. So that's a whole separate point that needs to be  
2 added.

3 DR. HANS: I'll wait for Sarah to get back down. Where I thought you  
4 were going, there are sort of two directions. One is to encourage, promote, and raise with the  
5 Secretary and the departmental leaders to incorporate the sort of concepts of genomics and  
6 genetics into a variety of ongoing activities.

7 The other direction which I thought you were going there for awhile, and  
8 now I have lost track of the numbers there, but is to say something like recasting one of the  
9 numbers that is up there, direct the HHS agencies to work together, and with federal, state, and  
10 private partners to, and I don't remember what words we used, collect all of the genomics  
11 education programs, and I don't remember where we were. And then work again with these  
12 same partners to promote the use of those materials through the various avenues that they have.

13 If you wanted to encourage HHS to go farther, even then just within HHS  
14 agencies, you could even ask that it be done in an interagency setting, like through the Quality  
15 Interagency Task Force that exists, that is a forum for interagency cooperation.

16 The second step is once you have sort of the collection in place, and you  
17 have sort of made the federal statement that this is what we should be doing, you then ask all  
18 the agencies to go back and look within their own programs, within their own regs, within their  
19 own activities, how do you appropriately incorporate it into what they're doing? Which  
20 certainly for VA and DOD, for instance, who run health programs, you have a more direct  
21 impact than you do just putting out recommendations to health professionals. So I'm sorry,  
22 Sarah, you probably weren't able to capture all of that.

23 MS. CARR: Sum that up.

24 DR. HANS: I was really looking down there where you have the red  
25 Number 1. So instead of framing it, because I thought that is where the discussion really was,  
26 instead of framing it as engage other health professionals, but rather to say direct HHS  
27 agencies, or direct the HHS to work among themselves, and with the state, federal, and private  
28 organizations that you have listed further down, to not facilitate, but actually catalog and  
29 disseminate genomics applications, and then work together to implement, where appropriate,  
30 within ongoing activities, or ongoing programs. We can work on wordsmithing that, but I don't  
31 know if you want to go that far, to be that direct.

32 DR. LEONARD: My concern is that I think we're losing the spirit of what  
33 is now the red Number 1. What the educational groups were asking for were cases that are  
34 relevant to everyday practice, so that they could do  
35 case-based education. This has now morphed into something that is not getting at, because it  
36 wasn't clearly stated, I think, in this one to begin with, that what you were looking for were  
37 cases that you could use for educational purposes, for case-based teaching.

38 So now that whole case-based teaching, the need to get the cases that are  
39 out there in everyday practice today, has been lost from this list.

40 DR. REEDE: I'm wondering if that is not a separate issue in terms of  
41 support for programs that will develop and help disseminate these specific cases, or interactive  
42 learning modules, or whatever that might be.

43 DR. LEONARD: Right, but it is now not in this list anymore.

44 DR. REEDE: I'm wondering if that shouldn't just be listed as separate. This  
45 list is going off in another direction, so I'm wondering if this is a separate entity.

1 Two other things, and given our 10-minute time frame that is just about  
2 used up, if we look at some of the other recommendations, there was one with regard to what  
3 was Number 5, "encourage accrediting, licensure, and certification of health professionals,"  
4 that that be changed. That was one of Debra's recommendations.

5 DR. LEONARD: It would be of health care and public health professionals.

6 DR. REEDE: Professionals.

7 DR. LEONARD: So certification bodies for health care. So up in the first  
8 line, Sarah, bodies for health care and public health professionals.

9 DR. REEDE: With regard to what was Number 6, a specific mention was  
10 made for us to keep in mind that cultural diversity and competency include the disability  
11 community. So I would question whether or not you'd like to add specific wording with  
12 reference to the disability community for Number 6.

13 Another item that we actually don't have time to word smith right now, but  
14 it was a strong point, was to have a recommendation that related to the ethical, legal, and social  
15 issues in training.

16 DR. McCABE: Any other big issues?

17 MS. HARRISON: I just had one about Debra's point about the public. I see  
18 that Sarah is trying to word smith something there to represent the thought. I just wanted to  
19 point out that Joe McInerney in his public comments did provide some wording for that, which  
20 I thought was pretty strong. So maybe that should at least be considered. One thing I wanted to  
21 add to it was to say that the initiatives should be done in a culturally sensitive and appropriate  
22 manner, so that we know that the initiatives that are put out there, that it is encouraged that  
23 some of them can be focused on certain communities that may otherwise not respond to other  
24 types of education.

25 DR. REEDE: Thank you.

26 DR. McCABE: Any other big issues?

27 (No response.)

28 DR. McCABE: If not, Sarah, we can try and work on these tonight and  
29 bring them back. But I think we do want to move on, so that we don't shortchange the public.  
30 What we're going to do now is take a three to five-minute break. Please don't go far. We are  
31 really just trying to link back up with Reed Tuckson. So please try not to go far.

32 (Recess.)

33 DR. McCABE: Before we proceed to the public comment portion of the  
34 meeting, I'd like to turn to Dr. Reed Tuckson, who is with us by phone -- that is sort of a  
35 hopeful statement -- to provide a brief summary of the inaugural meeting of the Advisory  
36 Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, in which  
37 he participated last week.

38 This congressionally-mandated committee is charged with providing advice  
39 and recommendations to the Secretary of Health and Human Services concerning grants and  
40 projects authorized under Section 1109 of the Public Health Service Act and technical  
41 information to develop policies and priorities for this program that will enhance the ability of  
42 the state and local health agencies to provide for newborn and child screening, counseling, and  
43 health care services for newborns and children having, or at risk, for heritable disorders.

44 Given the mutual interest in genetics, and the relevance of our committees  
45 to one another, Reed serves as a non-voting liaison to the advisory committee.



1 I would remind everyone that the largest body of genetic testing that is done  
2 in the United States is through newborn screening with 4 million babies born each year, and  
3 each one of them tested for between 3 and 30 disorders.

4 So with that as a preamble, Reed, I hope we can proceed.

5 DR. TUCKSON: Well, I hope that you can hear me.

6 DR. McCABE: Yes.

7 DR. TUCKSON: I appreciate the opportunity to report on the Advisory  
8 Committee on Heritable Disorders and Genetic Diseases. It was ably chaired by Dr. Rodney  
9 Howell, who is well known to many of us, and Michele Puryear, who has been involved with  
10 our committee, was one of the principal staff people, and she has done a great job.

11 You have introduced the mission of that committee, which is that the  
12 advisory committee is charged with providing technical information to the Secretary for the  
13 development of policies and priorities for the administration of grants for newborn screening  
14 and related programs and providing recommendations, advice, or information as may be  
15 necessary to enhance, expand, or improve the ability of the Secretary to reduce mortality or  
16 morbidity in newborns and children from heritable disorders.

17 During its first meeting last week, after much testimony from federal  
18 agencies and others, we heard a lot about the state of the art and the challenges that are  
19 involved in the genetic basis of newborn screening. As a result, the committee decided to focus  
20 on a few key areas in which we could make a significant impact.

21 First, the preliminary goals include making recommendations for a uniform  
22 panel of conditions for newborn screening, assessing the capacity needs of states' newborn  
23 screening programs and how they should be able to implement those, and making  
24 recommendations on parent education, notification, and informed decisionmaking.

25 This recommendation was also because there is an extraordinary amount of  
26 variability in the way individual states either require, mandate, or conduct newborn screening  
27 activities. It is not based upon an understanding of the best science in genetic screening, it is  
28 not based on a consistent application of that science, and it is differential in terms of the ability  
29 of states to collect information for long-term follow-up.

30 The committee also plans to study newborn screening tests and use the  
31 product of their work to develop recommendations on a nationwide core set of tests. This work  
32 will refine and extend a study that HRSA commissioned with Mike Watson and his team at the  
33 American College of Medical Genetics to carry out, which is sort of a landmark work in this  
34 area. The committee also plans to look at the costs of individual genetic tests and the costs of  
35 tests at the state level, including administrative costs. Finally in this area, the committee will  
36 carefully consider state-specific information such as workforce needs, the ability to assess and  
37 translate technologies into practice, and public education.

38 In addition, recognizing that the addition of new screening tests is likely to  
39 accelerate, the committee plans to develop a system or a process by which it will consider  
40 whether and how these new tests should be integrated and incorporated into the diagnostic  
41 panoply available for newborn screening.

42 Finally, the committee is interested in research needs to improve newborn  
43 screening and will use the American College of Medical Genetics study to assist them in  
44 recommending gaps in knowledge that would then focus on priority research initiatives and  
45 needs. They're also going to work carefully on creating a working group on interagency

1 coordination.

2 The only other point that I would make is that the committee was very  
3 impressed by our committee's strategic planning effort where we engaged in that process of  
4 identifying priority initiatives that require immediate and long-term action. They were  
5 interested in how we accomplished that task, and they have been given information regarding  
6 the process and lessons learned that we have from our experience. So I thought that was very  
7 useful that our committee's innovations have now spread to others.

8 With that, let me stop. It was a very good first meeting, and I am  
9 encouraged by the direction that this committee is setting off on.

10 DR. McCABE: Thank you, Reed.

11 Any comments on the report? Emily?

12 DR. WINN-DEEN: I just want to know if there is a written summary that  
13 either you have, or will be provided by the people who ran the meeting, so that we could get  
14 that sort of for our background reading?

15 DR. TUCKSON: Terrific idea. There will be a summary, and I'm sure that  
16 that will be easily made available to you.

17 DR. McCABE: Any other questions or comments?

18 (No response.)

19 DR. McCABE: If not, Reed, we know we are ably represented on that  
20 committee, and thank you for doing that.

21 DR. TUCKSON: Well, I also must say that, as always, I had terrific  
22 support from our staff. Our staff was there and is always great.

23 DR. McCABE: Thank you very much. Will you be able to stay with us?

24 DR. TUCKSON: Yes.

25 DR. McCABE: Thank you.

26 With that, we now move onto the public comment. One of our critical  
27 functions is to serve as a public forum for deliberations on the broad range of human health and  
28 social issues raised by the development and use of genetic technologies. So we greatly value  
29 the input we receive from the public. We set aside time each day of our meeting to hear from  
30 the public, and we welcome and appreciate the views that you all share with us.

31 We also have received written comments that can be found in everyone's  
32 table folders. I would especially like to call your attention to several requests that additional  
33 time be provided to allow the public to comment on the draft documents being considered at  
34 this meeting.

35 In the interest of time, I ask our commentators to please keep your remarks to  
36 five minutes, if at all possible. Today we will be hearing from -- and I'll give this in order so  
37 you'll know when you're up -- Kathleen Rand Reed from the Rand Reed Group, Andrew  
38 Fawcett from the American Board of Genetic Counseling, and Barbara Handelin from Handelin  
39 Associates.

40 First, Kathleen. So please come up to the table, as Kathleen is doing.

41 MS. REED: Good afternoon. First of all, let me say thank you very much  
42 to Dr. McCabe for just a couple of more extra minutes, and I want to bring two issues to the  
43 record, and to the table.

44 I followed the bouncing ball, and that is I stuck to the priorities, and to the  
45 determinations such as overarching issues, and the priority short term, and the highest priority

1 requiring in-depth study. The two comments that I would like to bring to the table, the first one  
2 would be continuing on genetic education and training of health professionals short term.

3 I am an anthropologist and an ethnomarketer, and I wanted to look at  
4 genetic issues that typically come into communities of color, especially those where the  
5 communities are what is known as hypersegregated. In other words, they are more than 95  
6 percent of that particular group, because oftentimes in those particular communities, it is a little  
7 bit more difficult to get information and ask some of the questions. And yet, if they are to be  
8 served as well, then those are the kinds of communities to look at.

9 First of all, let me also say thank you to Joe McInerney, who was gracious  
10 enough in January to allow me to present a poster for the NCHPEG meeting. The title was a  
11 little bit controversial, but it was called "U.S. Prison Policies, the Baby Daddy, and Genetics in  
12 the Hood." Let me tell you the key components of the poster, and I think you'll understand why  
13 I said it is controversial, but it is critical that these issues are addressed.

14 The key components for the poster were one, the removal and incarceration  
15 of young black males from hypersegregated intercity neighborhoods and some rural towns, the  
16 high incidence of teenage mortality from violence, poverty, and poor health, the resulting  
17 skewing of the male to female ratio, the phenomena of multiple matings, which often result in  
18 high consanguinity levels, i.e. the baby daddy.

19 A review of cohorts born during the height of the war on drugs in 1990 to  
20 1995, are currently on average reaching puberty in the years 2003 to 2008. The matings in  
21 many of these cases, because we have a member of the community that is often missing now,  
22 which is the gatekeeper, the genealogical gatekeeper, which is usually little old ladies that used  
23 to know who everybody was, and who belonged to whom.

24 Some of these matings may be between half-sibs, whose familial  
25 relationships may not be known within the general community. The potential is for possible  
26 epidemics of an increase in autosomal recessive disorders in this population, as well as a  
27 "blame the victim" mentality, and possible genetic redlining.

28 The reason I bring it to this particular group is because part of the  
29 comments that I got from the head of some of the genetic organizations, especially state-run,  
30 were chilling. They were chilling in this respect. As much as I was told that I was right, and in  
31 fact, I even had a deep conversation with Barbara Willis Harrison to get a reality check, to see  
32 if what I was thinking was absolutely true.

33 Ms. Harrison told me yes, we see this when I'm out doing pedigrees, this is  
34 a reality. And yet, one of the problems that we run into is this issue of genetics and the whole  
35 coming to terms with the community, and PC. What I found when I presented this poster, and  
36 I've had conversations with Mr. McInerney as well, is that many of the genetic counselors said  
37 yes, no, we see this in the pedigrees. The problem is because we are white, we can't do the PC  
38 part, because some people might think it is eugenics.

39 Now, I'm bringing these controversial issues. Anyone that knows me knows  
40 that I'm pretty much the one that always tries to bring up these kinds of issues, because some  
41 people don't, and I'm very comfortable doing this. But I would say that one of the things that  
42 we need to really work on, and this is an emergent situation, because I also went to Chicago,  
43 Detroit, Oakland, and Philadelphia and checked in and found that this is also a real big issue  
44 there, in many of the urban areas, and it is emerging.

45 I would say that we need to get on this and take a look. But one of the

1 things that we need to do is create these bridges where we have deep conversations and issues  
2 around who is white, eugenics, and PC issues. As they say, get over it. We need to have deep  
3 conversations so that we can protect these communities. Let me move quickly, because I don't  
4 want to run out of time, but I throw that out there as an issue.

5 The second one is, and very quickly, you have already identified the diverse  
6 representation of the population in clinical trials has particular importance in genetic research,  
7 since genetic variation among populations may account for differences in disease prevalence,  
8 drug reactions, and susceptibility to environmental triggers, among others.

9 There is a problem. The problem is I just came back from California,  
10 spending time going back and forth and being an expert witness in several cases where  
11 minorities are now suing. They are becoming more sophisticated about genetics, and suing  
12 because they are having serious adverse events with regard to their pharmacogenetic reactions  
13 to various prescription drugs and their respective diseases.

14 They are beginning to sue both the institutions, the hospitals, the HMOs,  
15 their physicians, and the pharmaceutical manufacturers. In the case of minorities, often their  
16 primary care physicians are also minority physicians, and often these physicians rely on the  
17 details and warrants issued to them from the pharmaceutical manufacturers for their prescribing  
18 guidelines.

19 There is now a scientific gap, and a legal gap, that causes injury to both the  
20 minority patient, and in some cases, the minority physician. When a lawsuit of this type is  
21 filed, all parties are sued. The hospital, the HMO, the patients, physicians, and the  
22 pharmaceutical manufacturer. But the pharmaceutical manufacturers, however, have an  
23 automatic defense, namely the learned intermediary doctrine, or the LID.

24 The LID can best be described as the manufacturer owes no duty to warrant  
25 to the patient, but to the learned intermediary, i.e. the prescribing physician. The liability, if  
26 anything, rests with the physician, or the institution, for the prescription to the patient.

27 However, this learned intermediary doctrine has several caveats. Namely,  
28 the information given to the physician must be "adequate." You as a committee have already  
29 identified the the flaw in this process, where you say pharmacogenomics, page 2, paragraph 3.  
30 "The pharmaceutical industry has very little incentive to do pharmacogenomic studies on  
31 "already marketed drugs or generic drugs." Such studies are expensive, appear to  
32 offer no market advantage for the sponsor of the studies, and the identification of persons for  
33 whom the drugs would be ineffective, thereby creating a stratified market for their products."

34 This disconnect causes several downstream consequences. Number one, it  
35 relieves the drug manufacturers of a look back, and the action with current information on  
36 pharmacogenomic information to offer not only adequate information to the learned  
37 intermediary, or the physician and the institution, but it also does not allow for the development  
38 of new information where minority participants were not included, and need to be included, and  
39 they need to have that look back.

40 From an ELSI perspective, this disconnect between the marketed drugs and  
41 revisiting old data, increases the discriminatory aspects which already face minority physicians,  
42 i.e. HMO's, and managed care. But it further reduces their numbers in the ranks, and to sum it  
43 up, this may wind up giving a look see also into some health disparities.

44 I would ask that you review these relationships, because the same  
45 relationships that occur with the drugs are going to be the same kind of relationships that you're

1 going to experience when you look at pharmacogenomics and the genetic testing.

2 Thank you very much, and I hope that has been helpful.

3 DR. McCABE: Thank you very much, Dr. Reed.

4 Are there questions or comments?

5 MS. REED: That's Ms. Reed. Not quite doctor yet.

6 DR. McCABE: Questions or comments? Debra?

7 DR. LEONARD: So this type of look back is being done in Japan, where  
8 they are asking all pharmacogenetic analysis for drugs on the market be done for the Japanese  
9 population. Would you suggest that the federal government ask for this type of thing to be  
10 done in all the ethnic populations? Because it is not just African Americans, but Hispanics, and  
11 all the different ethnic groups in the U.S., and that would be a major effort, because we are so  
12 ethnically diverse.

13 MS. REED: I think it is the old expression, how do you eat an elephant?  
14 One bite at a time. I would think to go up against this, as you say, straight up, would be a  
15 problem. One of the things that I'm making a recommendation on, I sat on an IRB for Heart,  
16 Lung, and Blood at NIH, and one of the recommendations is that there is an easier way to do  
17 this.

18 When Hopkins had a problem with the literature search, and now people do  
19 real deep literature searches, because no one wants to be shut down anymore. There is so much  
20 of a proliferation of information now, until one of the things that I'm asking is that when you do  
21 your literature search, that you run out as one of your key words, the ethnic groups, just to get  
22 the information back and see if there is anything out there, and then you can incorporate that  
23 into your research design.

24 But I do think that there is a responsibility for some of the older drugs that  
25 have been out there. One on the new end to put the responsibility on the pharmaceutical  
26 manufacturer to do that kind of inclusive work, which they should be doing anyway, but to also  
27 on the older drugs, to begin to revamp and take a slow approach, but still take the responsibility  
28 of a look back.

29 DR. LEONARD: Maybe this could come, Joe Hackett, through the FDA  
30 with some sort of truth in labeling, such that for different medications, it has to be stated what  
31 ethnic populations it has been studied in, so that at least as part of what comes with a drug, you  
32 know that it has only been studied in Caucasians, or Caucasians and African Americans, or  
33 something like that, so that physicians have some ability to know the adverse reactions.  
34 They may see adverse reactions in other populations in which the drug has not been tested.

35 MS. REED: And you are getting some of that in the DTC, in the direct-to-  
36 consumer advertising.

37 DR. HACKETT: My own perception is when you say that drug companies  
38 are very reluctant to do this, and we are trying to work with them on the newer drugs, it is all  
39 voluntary at this point. I can say again this year that there won't be any new diagnostic tests in  
40 using genomics this year, so it is slow.

41 MS. REED: At least it is on the table for you to examine. Thank you again.

42 DR. McCABE: Thank you, and I would point out that as we think about  
43 different ethnic communities, we need to think about genders, too, because that has been a  
44 problem in testing drugs as well. Thank you very much, Kathleen, for bringing that to our  
45 attention.

1                   Next is Andy Fawcett from the American Board of Genetic Counseling.  
2     Andy?

3                   MR. FAWCETT: I'm here today as a member of the Board of Directors of  
4     the American Board of Genetic Counseling. I'd like to begin by thanking the committee for  
5     allowing us this opportunity to provide comment.

6                   The ABGC is a national accrediting and credentialing body of the  
7     profession of genetic counseling. The ABGC establishes minimum requirements for graduate  
8     programs in genetic counseling, and develops a criteria by which individuals become eligible to  
9     sit for the certification examination.

10                  As this committee is well aware, genetics is one of the most rapidly  
11     advancing areas of scientific research, with clinical applications in practically all areas of  
12     medicine. The advances in medical genetics are forcing fundamental changes in the way health  
13     care providers practice medicine and think about health and disease.

14                  Knowledge about genetics and its social and ethical implications is  
15     becoming increasingly essential for many health care professionals, and must become an  
16     integral part of their curriculum. The draft resolution on genetics education and training of  
17     health care professionals developed by this committee clearly recognizes this need, and makes  
18     several solid recommendations related to integrating genomic concepts into our health care  
19     system.

20                  The ABGC, however, feels that it is critical that a distinct and separate  
21     focus on the education and training of genetics health care professionals be included in this  
22     resolution. As certified genetics professionals, we recognize that the demand for genetic  
23     counseling services will continue to increase, and unless more genetic counselors are trained,  
24     there will not be enough counselors to provide care to all patients and their families.

25                  Many certified genetic counselors devote significant time and effort  
26     educating other health care providers. The primary objective of these endeavors is to teach  
27     these providers to recognize a genetic condition in a patient, handle more straightforward  
28     genetic issues, and develop a relationship with genetic professionals in their community as a  
29     resource upon which they may draw to provide optimal care to the patient.

30                  This goal is not to teach other health care professionals to provide  
31     comprehensive genetic services. The issues surrounding genetic conditions are frequently  
32     complex, and as a result, a team of genetics professionals representing multiple genetic  
33     specialties may be required to provide this type of service.

34                  For example, genetic counselors have unique and extensive training in  
35     human genetics and counseling skills. They take didactic course work in human, medical, and  
36     clinical genetics, counseling theories and techniques, bereavement, crisis intervention, cultural  
37     competency, social, ethical and legal issues related to the delivery of genetic services, health  
38     care delivery systems, principles of public health, teaching skills, and research methodology.

39                  Genetic counseling programs accredited by the ABGC also provide  
40     extensive clinical training involving over 800 hours of field work, teaching, laboratory  
41     experience, as well as research. Genetic counselors are taught to prepare, deliver, and evaluate  
42     educational programs as they apply to various groups of learners. They are specifically  
43     qualified to deal not only with the complicated genetic and technical issues that often arise in  
44     the context of genetic evaluation counseling, but also the complex psychosocial, ethical, and  
45     legal issues with which patients and their families struggle.

1 Adequate financial resources must be in place so that an adequate number  
2 of individuals can be trained as genetic specialists. While the ABGC recognizes that  
3 educational efforts related to genetics must be aimed at our entire health care system, we  
4 encourage this committee to acknowledge that equally important is support for the continued  
5 existence of health care professionals who have specialized training in genetics.

6 Genetics health care professionals, such as genetic counselors, have been,  
7 and will continue to be, the ones who train and educate other health care professionals about  
8 the many complexities of genomic medicine.

9 Seventy-seven percent of all genetic counselors are currently involved in the genetics education  
10 of physicians and medical students, according to a professional status survey administered by  
11 the National Society of Genetic Counselors in 2002.

12 One-third reported teaching genetics to other health care professionals.  
13 Many genetic counselors have developed and implemented innovative educational models that  
14 facilitate the genetics education of other health care professionals and students. They are also  
15 involved in forming public policy related to genetics, and actively participate in genetics  
16 education programs for the public.

17 The recommendations made by this committee to ensure that genetics  
18 education and training of all health care professionals is adequate, will only be successful if  
19 there is an adequate genetics workforce to implement these recommendations.

20 Currently, there are 1,811 certified genetic counselors, and 25 accredited  
21 genetic counseling programs, and graduate programs in the United States, and three programs  
22 in Canada. Of those who responded to the professional status survey referred to earlier, only 6  
23 percent indicated they were non-Caucasian.

24 If genomic medicine is going to be equally accessible and practice in a  
25 culturally sensitive manner, individuals from minority populations must be recruited into the  
26 genetic counseling profession, and programs must be established in less populated areas of the  
27 United States.

28 In October of 2003, Robin Bennett, past president of the NSGC, presented  
29 data to this committee related to increasing the number and diversity of genetic counselors. To  
30 promote the training of genetic counselors, Ms. Bennett recommended that funding and  
31 granting opportunities be made available to support students and faculty in current genetic  
32 counseling programs, as well as to promote the establishment of new training programs.

33 The ABGC strongly supports the allocations of funds for this purpose if we  
34 are going to be successful in increasing the number and diversity of genetic counselors.  
35 Genetics health care professionals must reach out to other health care professionals to help  
36 them learn about the great promise and potential pitfalls of genomic medicine.

37 For this to be accomplished, we must ensure that there are competently  
38 trained genetics professionals. We strongly urge this committee to make specific and distinct  
39 recommendations to support the continued training of individuals in the field of genetic  
40 counseling, encourage increased diversity in our profession, and encourage those uniquely  
41 trained individuals to demonstrate their competency through certification and licensure.

42 Thank you.

43 DR. McCABE: Thank you, Andy.

44 Questions or comments for Mr. Fawcett? Yes, Debra?

45 DR. LEONARD: I think that this is an excellent point. We have often

1 discussed in this committee the paucity of genetic counselors, and so I think this is sort of an  
2 oversight in our education resolution, and we may want to consider addressing this in some  
3 way.

4 DR. McCABE: Thank you.

5 Any other comments or questions?

6 (No response.)

7 DR. McCABE: Next we have Dr. Barbara Handelin from Handelin  
8 Associates.

9 DR. HANDELIN: Thank you, Dr. McCabe and members of the committee  
10 for allowing this additional opportunity to provide some comments on your excellent  
11 deliberations, resolutions, and recommendations.

12 Let me just begin by saying that I'm here today principally representing  
13 myself as a Ph.D. geneticist who has been a technology developer and early practitioner of the  
14 provision of laboratory services and genetics in the commercial sector, as a consultant to large  
15 and small companies, and academic centers in the development of new genetic technologies  
16 and their implementation and practice.

17 I also speak from some 15 years of working with the IRB community in  
18 improving the oversight of genetics research involving human subjects, especially those that  
19 were sponsored by private industry.

20 I provided some written comments. I'm not going to reread those to you  
21 here today, but rather to move on to some additional thoughts that I have had in the meantime,  
22 in particular influenced by your conversations this morning.

23 I wanted to respond to the question that Brad Margus raised earlier about  
24 what may be broken, or is there something broken in the practice of medicine today with  
25 regards to genetics inclusion? I had the following thoughts.

26 While I'm routinely amazed by how often the genetic basis of a set of  
27 symptoms is not considered in a differential diagnosis, I also have to admit that I have an equal  
28 number of stories of people who have spent years of suffering and expense, failing to get a  
29 correct diagnoses, even when genetics is not involved. I know that all of us do.

30 I think that this is a reflection of the way we are perhaps not teaching  
31 physicians to think about a constellation of symptoms, about how to ask enough questions and  
32 hear enough answers about a patient's condition. But most importantly, how to think through a  
33 complex problem, considering many alternative diagnoses, as well as the physician being able  
34 to think about what is all the data that I'm going to need to collect in order to weave amongst  
35 those possible alternatives.

36 I would posit that perhaps this also says, most importantly, that we have far  
37 exceeded the capacity of the majority of physicians to keep a reasonably complete fund of  
38 knowledge of human disease in mind as they are seeing patients. Genetics are not.

39 In this vein, I would strongly support the idea that arose earlier today, I  
40 think originally by Dr. Tuckson, and then echoed by Sarah Carr about the timeliness of  
41 promoting the development and implementation of information technology systems that can  
42 support routine clinical practice, including the suggestion of genetic etiologies or contributions  
43 to common disease in everyday practice.

44 But I make this suggestion with a caution, and that being that these sort of  
45 practice tools have been developed previously and marketed by private companies, only to be



1 rejected by many physicians because they don't like the feeling that a computer may appear to  
2 be doing my job. That kind of sense of pride in providing knowledge and experience is a  
3 barrier to the acceptance for such tools, and it causes me to wonder if perhaps we need to see  
4 some kind of requirement for such tools in large medical practices, in large institutions, or the  
5 requirement for such tools to be incorporated into new training programs.

6 So I'd also like to speak to several other points that were contained in the  
7 various briefs that the committee provides as background. In particular, I'd like to speak to the  
8 brief on large population studies. Genetics as a discipline, I learned when I was a Ph.D.  
9 student, is not the study of inherited traits, nor the study of inheritance itself, but rather more  
10 fundamentally, the study of variability. Variability in populations is sourced through inborn  
11 traits of a species. Variability in populations is the source of our biochemical patterns, our  
12 physiology, our response to infectious disease, to nutrition, in the form of allergies, for  
13 example, to exposure to xenobiologicals, plant toxins and venoms, to daily life stress, and to  
14 administer treatments, as in the case of pharmacogenetics.

15 Variability in populations is the source of our racial and other group traits,  
16 as we share specific variants with family members, and choose parenting partners within our  
17 close at hand community groups. Variability in populations allows us to be at once a part of a  
18 group or sector, and to be individuals.

19 I would posit to the committee that the basic goals of our pursuit of  
20 knowledge about the human genome is fundamentally about understanding the variability in  
21 populations. Therefore, ensuring the potential to undertake such large population studies all  
22 across the United States through the various strata of our diverse population seems to me to be  
23 among the most important missions of all persons and committees who are charged with  
24 overseeing the exploration and application of genetic technologies in research and medicine.

25 So I hope that you here, and those at the NIH, and the NHGRI, for example,  
26 will focus on finding ways to list the main barriers to the ethical and productive conduct of  
27 large studies. I agree wholeheartedly with the observation made in the committee brief, that  
28 "the lack of the universal health care system, and the lack of a uniform electronic medical  
29 record system here in the U.S. are likely two very important barriers today."

30 The simple promise of guaranteed health insurance and health coverage, as  
31 are seen in other countries in the world, such as Sweden and Iceland, go a long way to  
32 encouraging people to participate in such large research protocols.

33 Other safety concerns for us is subjects, of course, about privacy and  
34 confidentiality. I would suggest that it has been now my conclusion I guess that we cannot ever  
35 guarantee absolute privacy, or that we could hold out all individual details completely  
36 confidential. Therefore, it seems paramount to me that we must feel confident that those who  
37 may choose to attempt to harm us through use of our private information will not be tolerated,  
38 that there will be severe consequences for such abuse of privacy.

39 I have an uncle who is an attorney who used to say that no one would cheat  
40 on their taxes if the death penalty were the required sentence. And so to be clear, while  
41 providing clear requirements for utilizing all the available methods for shielding private  
42 information from wrongful use, I believe it is equally important to develop a system of clear  
43 consequences for such wrongful acts, to send a message that as a society, we will not tolerate  
44 such violations of respect for persons.

45 In conclusion, I just want to say that I am using this language of respect for

1 persons, which I take from the Belmont Report, one of the three general principles laid out by  
2 the Belmont Committee in the Protection for Research Subjects as a way to remind ourselves,  
3 again, that the reason that we have rules and regulations about the ethical use of human subjects  
4 arises from the misuse of genetic and other kinds of information here in the United States, and  
5 in Germany decades ago.

6 So I urge us to return to those simple principles, respect for persons,  
7 beneficence, and justice, as you continue in your important work here. The final point that I  
8 would like to take a minute to make is to applaud you on your thorough and thoughtful  
9 treatment on the reimbursement and coverage issues for new technologies, including those in  
10 genetics.

11 Since the first application of our knowledge of genes and performing carrier  
12 tests and prenatal predictive tests for single-gene disorders, we have been practicing a new kind  
13 of preventive medicine. I suggest that today, our nearest benefit from the genome knowledge  
14 base remains primarily in preventive medicine. As we see the very first of new cancer  
15 screening tests, first tests to identify patients who will or will not benefit from toxic  
16 chemotherapies, these are the kinds of new products that I think are going to typify the  
17 implementation of genetics in the next decade.

18 Therefore, I would especially encourage you to drive home the importance  
19 of creating a sea change in our coverage for preventive medical services, so that we can  
20 encourage to participate in the research necessary to get there, as well as in the enjoyment of  
21 those benefits.

22 Thank you very much.

23 DR. McCABE: Thank you, Dr. Handelin.

24 Any questions or comments, anyone?

25 (No response.)

26 DR. McCABE: Thank you very much.

27 I'm sorry. Emily.

28 DR. WINN-DEEN: I just had a question. How much do you think of the  
29 reason that genetics isn't considered is because physicians in their time constrained, by the  
30 clock office visits, are basically applying the 80/20 rule, where you take the most common  
31 diagnoses and sort of farm everybody through those, and just don't really have the time to  
32 consider the less likely scenarios, or the more infrequent scenarios?

33 DR. HANDELIN: Well, I guess I would question whether genetic  
34 etiologies, or genetic components of many common diseases are typically the uncommon  
35 explanations for many presenting symptoms. But I agree with you that certainly a reluctance to  
36 consider many other alternative diagnoses clearly falls principally from a lack of time right  
37 there in the office, as well as a lack of time to incorporate new ideas, new concepts, and new  
38 knowledge into your fund of knowledge. Again, I think it is pointing to the need for some  
39 assistance in doing that.

40 DR. McCABE: Any other questions or comments?

41 (No response.)

42 DR. McCABE: Thank you, Dr. Handelin.

43 The way we will proceed now is we're going to have Cindy Berry give her  
44 presentation, and then we'll take a break before the discussion of coverage and reimbursement.

45 At the March meeting, the committee decided that this topic, coverage and

1 reimbursement, would be the subject of its first major report. A draft report was prepared by  
2 staff, and can be found at Tab 5 of your briefing book.

3 I'd like to take this opportunity to thank Cindy Berry for her leadership on  
4 this issue, and Suzanne Goodwin, and Amanda Sarata for preparing the draft report. I think  
5 they have done a remarkable job in a very short amount of time. I think we should  
6 acknowledge, however, that this is a draft, and neither Cindy nor the staff have given it any  
7 "final blessing."

8 Also, as I indicated before, I doubt that we will complete the discussion of  
9 this topic here, but that it will proceed into the next meeting. So now at this time, Cindy is  
10 going to give us an overview of the scope and content of the issues covered.

11 Cindy?

12 MS. BERRY: Thank you, Ed.

13 In the interest of time, I'm not going to go into enormous detail on each of  
14 the topics that are covered under the report, but rather give a general overview of how the  
15 report is structured.

16 Many of you, Reed in particular, have looked at this in great detail and have  
17 provided some comments, and this afternoon's efforts are going to be focused on really taking a  
18 good, hard look at the report. So at this time, I'm not going to go over specific language. We  
19 all are going to have edits, comprehensive edits, I suspect, on some things that have been  
20 overlooked that we will want to make sure are included in the report, and there are things that  
21 may not be addressed adequately, and we'll need to flesh them out in greater detail. But I'll just  
22 give you a general feeling for how this report is structured, and then we can proceed to the  
23 discussion phase where we can get into some of the details.

24 As you may remember from the last meeting, we did have a pretty elaborate  
25 priority-setting process. Out of that process came coverage and reimbursement as one of the  
26 priority issues that this committee should look into, and coverage and reimbursement being one  
27 of the key barriers to access the genetic technologies.

28 The purpose and goals of the report really are to give an overview of the  
29 current state of play. What is going on right now in the area of coverage and reimbursement for  
30 genetic technologies and services, and how can we improve that?

31 The sections of the report are as follows. We have an introductory section  
32 of course. Genetic technologies and services, what are we talking about when we talk about  
33 genetic technologies? Reed has brought this up in the past, and I think we may need to do a  
34 little bit more work there as in particular, let's define this. What is the scope of genetic  
35 technologies and services that we're talking about? General principles.

36 Then in the background section, the report delves into our health care  
37 financing system in the United States. How are coverage decisions made in federal health care  
38 programs, and in the private sector? How are services and technologies being reimbursed?  
39 What is the billing process? And then after that general overview, the report delves into the  
40 barriers to coverage and reimbursement, and of course the recommendations section is  
41 something for this committee to come up with.

42 Basically the introduction sets out what the problem is that genetic  
43 technologies can help enhance clinical care and improve outcomes, but there are limitations in  
44 terms of what is covered, and what is reimbursed, and those are barriers.

45 This section on genetic technology, as I mentioned, goes into what are we

1 talking about when we talk about genetic technologies and services? What distinguishes them  
2 from other health care services and technologies?

3 The general principle section really has sort of an outline of some things  
4 that we have discussed in this committee in the past, and that undoubtedly we will delve into in  
5 greater detail in the future. It is not a comprehensive or extensive list, you will see others there  
6 at the bottom as we go through the discussion this afternoon, I think folks will come up with  
7 additional principles that probably need to be incorporated in this report.

8 As I mentioned, this is the general overview of our U.S. Health Care  
9 System, the different federal programs, the private sector programs, managed care, and then the  
10 issue of the uninsured, and underinsured. The section on coverage decisions reviews how  
11 Medicare makes coverage determinations, and then it goes into the private sector  
12 determinations, which really are varied. Every health plan is different, every health plan has  
13 different guidelines, and the results and outcomes are different.

14 The situation is really the same in terms of payment. In Medicare, you do  
15 have a system for determining what is reimbursed, and the report does go into what the current  
16 payment rates for genetic technologies and services are, and we'll undoubtedly have a  
17 discussion about whether those are sufficient. I suspect based on previous testimony and  
18 comments that we've heard, that payment is not sufficient for many of these services.

19 The report also goes into billing processes. Coding, CPT codes, and who is  
20 entitled to bill for genetic services and technologies. The next step, of course, is to then  
21 examine the specific barriers, barriers that are particular to the Medicare program, barriers to  
22 access that are particular to Medicaid and state programs, and then of course there are barriers  
23 that are applicable to all insurers, whether they are in the public or private sector.

24 We'll just run through a few of these very quickly. In Medicare, for  
25 example, the screening exclusion is one of the barriers that the report identifies. If there are no  
26 signs, symptoms, complaints, or personal history of a disease, and there is not an injury, it is  
27 going to be difficult, if not impossible, for Medicare to pay for a screening test. That, of  
28 course, is based on the Medicare law itself. It is not something that you can necessarily fault  
29 CMS for. They are going by what the statute dictates, and so this is a matter that we'll discuss  
30 later, is a change in the statute warranted.

31 The report goes into detail about local medical review policies and national  
32 coverage decisions. There is sort of a tension in some respects between local decisions as to  
33 what is covered, and then a national coverage decision. How do those interact, and how do  
34 they affect genetic services and genetic technologies?

35 Genetic counselors, are they able to submit bills to Medicare for their  
36 services? The answer is no, according to the statute. There are issues with regard to private  
37 health plans undoubtedly as well. Medicare is a national leader in health care. This is an  
38 interesting point, because in many, many cases, we see that Medicare really sets the stage and  
39 serves as a guide for other insurers.

40 So if Medicare covers something or doesn't cover something, often that is  
41 replicated in the private sector, but not always. My personal experience, and others will have  
42 experience here as well, when it comes to preventive services, oftentimes we see the private  
43 sector taking a more proactive role simply because the Medicare program is really bound by  
44 statute. As we all know, Congress doesn't move too quickly, and it is very difficult for a  
45 legislative body to keep up with advances in health care.

1           In this case, sometimes the private sector actually is able to move more  
2 quickly. So this will be a discussion in the report that explains why the Medicare program is so  
3 important, not only for the Medicare beneficiaries, but as a model for other insurers, but  
4 conversely, maybe there are some lessons that we can learn from the private sector in terms of  
5 their ability to incorporate some of these technologies in health care.

6           The report goes, as I mentioned, into state programs, and then we have  
7 barriers that are applicable to all insurers, and all health care programs. Medical necessity  
8 criterion, and of course one of the underlying issues, threshold issues that we will undoubtedly  
9 discuss in comments today has to do with the informational value of genetic tests, and genetic  
10 test results.

11           If it is just great for someone to know the result of the test, does that  
12 warrant coverage under a federal health plan, or private health plan. There is the issue of the  
13 future benefit, testing someone for a genetic disorder. If there is no treatment available right  
14 now, is that, again, an informational benefit? And also, if someone is not experiencing any  
15 signs or symptoms, if you test now, that will produce some benefit at some point in the future,  
16 is that a barrier to coverage, whether it is in the federal health programs, Medicare, or whether  
17 it is in the private sector?

18           Of course, the role of cost and coverage decisions. It is not explicitly  
19 considered in the coverage decisionmaking process, but undoubtedly it is a factor, and we'll  
20 have to be mindful of that.

21           Experimental exclusions. Some of these technologies fall under that  
22 category, and that's another barrier to coverage and reimbursement. CPT code modifiers, that's  
23 an important part of the report, because it outlines really the process for getting a CPT code,  
24 and what that means for coverage and reimbursement, and for payment. There are no CPT  
25 codes specific to genetic counseling, and that could be a barrier as well.

26           Evidence-based coverage decisions, we talked a little bit about that this  
27 morning. It is a current theme in our comments today, as well as what is going on in the report.  
28 Obviously a lot of insurers, rightly so, feel the need to have some kind of data with which to  
29 make a decision, and on which to make a decision to whether a treatment, test, or some  
30 technology or service should be covered.

31           We have talked a little bit about how important it is to have this kind of  
32 research available so that we can help make the case if certain technologies should be covered.  
33 Right now, there is a little bit of, and I think actually not a little bit, a pretty significant need for  
34 that kind of information.

35           The report goes into payment rates, and the issue of low reimbursement. It  
36 is not necessarily unique to genetic technologies and services. It is a common complaint, I  
37 think, from many providers of health care services. But the question is if the reimbursement is  
38 so low, and oftentimes is below the cost of delivering the service, or delivering the product or  
39 the test, what impact does that have on access?

40           There are broader issues that the report discusses. It doesn't attempt to  
41 resolve all of them, because these issues transcend genetics. They really go throughout the  
42 health care system, but we have talked a lot today about, and we will continue to discuss the  
43 issue of health disparities.

44           Education and training, we have spent a lot of time on that today. I think  
45 probably I should close with this, topics for discussion this afternoon. The overall structure of

1 the report, the content, the tone of the report, the general principles, and then we have to come  
2 up with some recommendations.

3 Now, I'll bring up a point. In one section of the report if you have read it,  
4 there are some boxes on policy options. My view, it might be a little bit confusing to have  
5 policy options there, and then have recommendations someplace else. We might want to figure  
6 out how we want to structure that. I don't know if others agree or disagree, but I think the  
7 charge for this committee is to finalize the report, make sure that we've captured all of the  
8 basics, and that the language is something that we all feel comfortable with, but really hone in  
9 on some of the recommendations, concrete recommendations, that this report can make to  
10 improve coverage and reimbursement for genetic technologies, thereby improving access to  
11 these services.

12 Here are some of the topics, and I'll leave this slide up for our discussion.  
13 These are topics that staff have identified that we could go into all of them, some of them, a  
14 few of them, and it would be important to get the thoughts from the folks on the committee as  
15 to priorities, issues that you want to focus on, and that you think should be the basis for our  
16 discussion today for purposes of putting together some concrete recommendations for the last  
17 part of the report.

18 DR. McCABE: Thank you, Cindy.

19 I think with that, why don't we take a 15-minute break. Again, we have the  
20 refreshments up here for the ex officios, the members of the committee. We'll reassemble here  
21 in 15 minutes, so around a quarter after.

22 (Recess.)

23 DR. McCABE: Let's begin again, please. I'd like to remind everyone that  
24 our goals for the next roughly two hours are to discuss the draft and develop recommendations  
25 to be included in this report to the Secretary. Again, I doubt that we will finalize this over the  
26 next two hours.

27 We have written public comments and have heard public testimony  
28 regarding the report. Cindy has identified some areas where we might want to focus our  
29 attention, so I will turn it over to you now, Cindy, to lead our discussion for the next two hours.  
30 Our plan is to finish up here at 5:30 tonight.

31 In case I forget, let me remind everyone that we are starting earlier  
32 tomorrow. We will start at 8:00 tomorrow morning. Thank you.

33 Cindy?

34 MS. BERRY: Based on the amount of time that it took us to go over the  
35 education and training resolution, it seems like, and here is just a suggestion, but let me know if  
36 anyone disagrees. That for specific edits, because the report is so extensive, perhaps we should  
37 give a deadline to ourselves to hand write, or email specific wording changes and things like  
38 that, and get them to staff, to Suzanne. I don't know what deadline you want to put on there,  
39 and focus our discussion today more on the bigger picture.

40 Well, the structure of this is wrong, or we need to add a section on such or  
41 such, or here are what our recommendations should be, that kind of a thing. I don't want to  
42 unilaterally impose that, so tell me if you disagree. But it seems like that might be a more  
43 efficient way to deal with this today.

44 DR. McCABE: Our next meeting is October 18th and 19th. We'd want to  
45 have time to get those well incorporated. When would be a reasonable time, Cindy and

1 Suzanne? A month, six weeks before that? Six weeks?

2 MS. BERRY: Where does that take us?

3 DR. McCABE: If it was six weeks, that would be the beginning of  
4 September.

5 MS. GOODWIN: What does this relate to?

6 DR. McCABE: To get in feedback on the document even sooner than that.  
7 Mid-August? Do I hear mid-August? Even earlier. Staff is much more reasonable about time  
8 than I am. So early August? Mid-July?

9 MS. GOODWIN: Mid-July, or even early July, depending on how  
10 extensive the editing process needs to be. Part of that relates to when, if we plan to go out for  
11 public comments, but in time for the October meeting. So there will have to be significant time  
12 to prepare for the Federal Register notice for public comment, and also to give the public  
13 sufficient time to respond. Also in the Federal Register notice to make sure that we publish, or  
14 have available, a revised draft, based on your edits and today's discussion.

15 MR. MILLER: How about Thursday?

16 (Laughter.)

17 DR. McCABE: Well, if we give people a month, that would be roughly the  
18 16th of July. If we want to do it even earlier than that, we could go for the 1st of July, which is  
19 a Thursday, as per Paul Miller. Okay. So July 1. So two weeks would be July 1. Would that  
20 work for staff?

21 MS. GOODWIN: That's good. Thank you.

22 DR. McCABE: Okay. So July 1 it is.

23 DR. PHURROUGH: For those of us who are representing other people, for  
24 those people that we're representing, do they get an electronic version of this?

25 MS. GOODWIN: There is a copy available, a PDF copy on our website.

26 DR. PHURROUGH: Okay.

27 MS. GOODWIN: There is also one that was sent electronically to you  
28 before the meeting, but we can certainly send it out again after the meeting to make sure that  
29 you have a copy.

30 MS. BERRY: Emily?

31 DR. WINN-DEEN: I was going to suggest that perhaps what we should do  
32 is rather than focusing, as you mention, on edits on sort of the background information where I  
33 think we've gotten some comments in that some of the background information needs updating,  
34 but I think it is very important for us to discuss the policy recommendations. I certainly would  
35 like to see, because this is going to be a topic that I think already has evoked a lot of public  
36 comment and is likely to evoke more public comment, that we try and discuss those policy  
37 recommendations in this committee.

38 But also I would like to make a suggestion that it be circulated one more  
39 time for review to the committee before it gets posted on any site anywhere, so that if we have  
40 additional comments on each others' comments, we can sort of get that all worked out ahead of  
41 time, if it is possible to do that.

42 I know I got a whole bunch of calls the day it went up on the website. What  
43 are you guys thinking about? How did you get to this, that, and the other thing? I'm like, gee, I  
44 haven't even read it yet, I don't know what we're thinking about. I think when we have  
45 subcommittees, we need to somehow share the subcommittee work with the whole committee,

1 at least a little bit of time frame before it is shared with the --

2 MS. BERRY: Well, in this case, we didn't have a subcommittee at all, this  
3 was a staff prepared document. Now, is there a requirement that it be posted? That's the  
4 question I have, is are we able to circulate it amongst ourselves and comment on it without it  
5 being posted until it is in final form? Or is there some sort of requirement for our committee  
6 that it needs to be posted?

7 MS. GOODWIN: The reason why this draft was posted before the meeting  
8 is, well, first of all, staff was working on it up until right when the briefing books were sent out.  
9 Part of the reason we opted to post it before the meeting was part of the FACA requirement,  
10 the Federal Advisory Committee Act, requires us to make documents that will be discussed in  
11 detail during the meeting, available to the public, so that they can participate in the discussion.

12 DR. WINN-DEEN: Yes. So I didn't have any problem with it being  
13 publicly posted. I just felt like we were caught a little bit flat without having something that  
14 went out, representing this was a work product of the committee, and the committee hadn't even  
15 seen it. That was all that my comment was directed at, that I'd just like to have a little bit of a  
16 time window for the committee to actually have read it and thoughtfully gone through it before  
17 we start getting a lot of feedback.

18 DR. McCABE: Yes, it does have to do with FACA, and our practice with  
19 respect to FACA. That was actually my decision, that we would do that, because we wanted  
20 the public to be aware of this before we came here, so that was my decision, and I'll take  
21 responsibility for that.

22 DR. REEDE: I think part of it is if you look at the top, on each one of them  
23 it says it is a staff draft, however, and that it didn't represent the views of this committee, or of  
24 the government. I don't think anybody reads the fine print, but the fine print did say that.

25 MS. GOODWIN: But certainly before the next meeting, there will be an  
26 opportunity to go back and forth as a committee by email to revise the next draft.

27 DR. WINN-DEEN: Thank you.

28 MS. BERRY: Does anyone have any suggestions for kicking off the  
29 discussion on policy recommendations? Are there issue areas that you want to prioritize for  
30 us? Steve?

31 DR. PHURROUGH: I'll make a few comments from the CMS viewpoint,  
32 since we are seeing it for part of the discussion. First of all, on the draft in general, a big  
33 concern in your first principle is we need to collect data, and yet that was very minimally  
34 discussed throughout the report, the importance of a coverage decision being evidence-based.

35 We sort of tied coverage and reimbursement together, and you really need  
36 to separate that concept. Certainly speaking for CMS, and I suspect for most private insurers,  
37 at least in reading Allan Korn's comments, and Reed's also, when we as payers make decisions  
38 about the kinds of things we're going to cover, we want to make sure that there is a good  
39 evidentiary base for it.

40 That evidentiary base, at least in the Medicare world, is there are good  
41 quality controlled trials that demonstrate that the application of this technology improves the  
42 outcomes of beneficiaries that we have responsibilities for.

43 It brings up a whole host of questions in genetic testing that we, as a  
44 society, and we at CMS, need society to have this answer. One is in some cases, genetic testing  
45 will alter treatment, but in many cases, it won't, it will just be information. I think that



1 something perhaps even broader than us is a question of whether health care dollars should be  
2 expended for information when it will not alter outcomes. We've had that discussion internally,  
3 and that is certainly a topic for further discussion.

4 More specific, policy issues, specifically around the screening exclusion.  
5 The draft is correct that it has been a longstanding Medicare policy that Congress prohibits us  
6 in its language from paying for tests that are screening in nature, or in most cases, paying for  
7 preventive services.

8 We have had a lot of discussion in the last several months as to whether in  
9 fact it was Congress' intention. There are some viewpoints that say that CMS could change its  
10 mind without congressional input. Even those who think that it could only done through  
11 rulemaking, but even those who think that may be possible strongly suggest that we should let  
12 Congress tell us to do that.

13 Since Congress has been very involved in adding screening and preventive  
14 benefits over the last decade, and they seem to enjoy that, and they have yet to give us a broad  
15 mandate to do screening and prevention, preferring to, how shall I say, receive the accolades of  
16 adding a new benefit that they may get from their constituents, which is in fact their job.

17 The agency could, based on our legal authority, perhaps change our  
18 screening exclusion, but we're not sure that that would be well received in Congress, and  
19 believe that the congressional direction is probably the preferable solution. And in fact, there is  
20 legislation in front of Congress now sponsored by a whole host of members on both sides of the  
21 aisle that I believe has been encouraged by the Partnership for Prevention, that would allow us  
22 to do coverage determinations on screening and preventive services, if they were in fact  
23 recognized as beneficial by a national organization.

24 So there is some legislation there. We are not a lobbying organization, but  
25 as we discussed on the education and training earlier, that may be a role for this committee to  
26 encourage.

27 On the other policy of who can and can't be paid, that is very clearly a  
28 congressional action. We can only pay people that Congress says we can pay. Congress has  
29 been very hesitant over the almost 40 years of Medicare to expand that list, having only  
30 expanded it a couple of times in that 40 years from physicians to one other occasion, nurse  
31 practitioners, CNSs and PAs and in very narrow instances, physical therapists, and most  
32 recently, a very narrow indication for dieticians.

33 That is going to be an extremely difficult barrier, I suspect. Congress is not  
34 very receptive to adding new providers. So that is the only way we could pay genetic  
35 counselors, and that is going to require some significant support from others outside this  
36 committee to encourage them to do that.

37 MS. BERRY: Steve, do you happen to know the bill, the legislation that  
38 you referred to? The sponsor of the bill number?

39 DR. PHURROUGH: I don't have that with me. I can see if I can locate  
40 that, and provide that to the staff.

41 MS. BERRY: Terrific. Does anyone else have any comments? Emily?

42 DR. WINN-DEEN: Well, so, in the great minds think alike kind of thing,  
43 one of my comments back was why can't we encourage Congress to enable groups like the  
44 preventive task force when those recommendations come by a "deemed body" that any  
45 recommendation that is coming from certain institute medicine or USPSTF, that those would be

1 automatically then covered prevention services.

2 I guess what I'd like to ask is so which way do you think the wind is  
3 blowing? And is this something, because CMS and CDC that is sort of running the prevention  
4 side of things, both report to our guy, who we are supposed to make recommendations to,  
5 Tommy Thompson, is it possible for us to send something up to Secretary Thompson that  
6 basically says, we think this would be a good thing? Or are there other political things that you  
7 should make us aware of?

8 DR. PHURROUGH: I'm not an expert in how the legislative process works.  
9 But as I understand it, Secretary Thompson provides to Congress every year what is called the  
10 legislative budget, which has nothing to do with money. It has to do with these are things that  
11 we recommend that you consider for enactment.

12 Someone will have to check and see if I'm correct, but I think that is the way  
13 the system works. We could encourage the Secretary to add that to that particular  
14 recommendation to Congress, which is not then us as government employees lobbying  
15 Congress directly, but using the vehicle that Congress has established, this legislative budget. I  
16 think that's the terminology.

17 DR. McCABE: Yes, I think that's an important point. The process is that  
18 we advise the Secretary. We can be specific as we have with genetic nondiscrimination with  
19 respect to influence on the legislative branch, but that's the mechanism. It is through Secretary  
20 Thompson.

21 DR. WINN-DEEN: Yes, I think my point was that since sort of both parts  
22 of this equation report to him, that it is definitely sort of 100 percent under his umbrella to be  
23 able to do something like that.

24 DR. TUCKSON: This is Reed. Can you all hear me?

25 MS. BERRY: Yes.

26 DR. TUCKSON: (Inaudible.)

27 DR. FELIX-AARON: That's right. AHRQ does do prevention as it applies  
28 to clinical practice, and what goes on between doctors and patients as distinct from sort of the  
29 community preventive services, which is under the purview of CDC.

30 Reed, are you finished? Because if you aren't, I'd like to make a point here,  
31 but I'll wait.

32 DR. TUCKSON: Go right ahead.

33 DR. FELIX-AARON: Right. I mean, in listening to both Steve and the  
34 conversation which went earlier, somebody mentioned the partnerships for prevention. The  
35 question, I hear us going around, is sort of what are the levers for prevention?

36 I think one we can do is sort of say, this is genetics, how do we push this  
37 forward in terms of looking at coverage for these specific services? What I hear is that genetic  
38 services are part of a larger category of preventive services. And so the question I think we're  
39 faced with is what are the levers for prevention, and who are the major stakeholders?

40 So I was very struck by your point, Cynthia, where you said that some of the  
41 innovators in this area is not necessarily the Medicare program, but actually private insurance  
42 companies, in that they are much more advanced in terms of prevention than Medicare. And so  
43 those groups are ahead of the pack, so to speak.

44 So the question for, or not the question, but a question for us is really trying  
45 to understand what is going on in prevention, and who is pushing prevention in this country,

1 and what can we as a group do to sort of piggyback our efforts either to supplement what they  
2 are doing, or to augment what is going on in those circles, and what advice for the Secretary,  
3 how can we provide the Secretary with guidance which strengthens what is going on in that  
4 arena?

5 MS. BERRY: Reed, you're probably the best person to address it. Just to  
6 put in context, my comment wasn't at all that CMS doesn't want to go down the road of  
7 prevention, but simply their hands are tied by the structure of the Medicare program, legislation  
8 in Congress, and all of that.

9 The private sector doesn't have as many of those constraints, and so they're  
10 a little bit more at liberty to be innovative, and to respond more quickly to innovations. But  
11 Reed, I'd defer to you. Do you care to address that point?

12 DR. TUCKSON: (Inaudible) how do we give the Secretary a reasonable  
13 body of work to get around? I think that the implicit guidelines are that we do want to see  
14 appropriate genetic prevention diagnostic services to be incorporated into the  
15 Medicaid/Medicare rules.

16 The issue is what does that mean? I think that (inaudible) automatically  
17 anytime there is a partnership or prevention (inaudible) there are some other issues here. I  
18 think what our challenge is, without having to (inaudible) that is the issue.

19 MS. BERRY: Reed, you cut out a little bit at the end where you were  
20 making the big point.

21 DR. TUCKSON: Well, (inaudible) responsible way, and those issues and  
22 those questions are no different for the public sector than they are for the private sector.

23 DR. WINN-DEEN: Reed, can I ask you a question from the private  
24 insurance point of view? How far do recommendations from things like the U.S. Preventive  
25 Services Task Force go in the private sector to encouraging coverage for services, just sort of in  
26 general?

27 DR. TUCKSON: They are exceedingly important. Because again, they  
28 provide the kind of very rigorous evidence-based analysis for (inaudible).

29 DR. PHURROUGH: I'm Steve Phurrough from CMS. One of the  
30 difficulties in waiting for the USPSTF is that their process is a very detailed, slow, arduous  
31 process, as it should be. I think based upon the amounts of evidences that are out there today,  
32 the benefits of genetic testing, that they're a long way from coming up with recommendations  
33 that if we were to be told to follow them, and aside, our preference would be we're given  
34 permission to use their recommendations versus told to follow them, but that's a different  
35 subject. I think they are a long way from coming up with those particular recommendations.

36 I think the real emphasis that we need to be encouraging is there are some  
37 policy changes that need to occur, but we need an evidentiary basis that's just not there, or is  
38 there to a very small extent, particularly in the populations that we are concerned with in the  
39 Medicare population.

40 There is a whole host of evidentiary basis for the younger populations, but  
41 I'm not sure that I need to worry about ordering a PKU for my population I'm concerned with.  
42 We need an evidentiary basis if Medicare is going to cover genetic testing, for that particular  
43 population, and the benefits of that testing in that population.

44 So I strongly agree with Reed's comments and his written comments, that  
45 that is the key factor that needs to be encouraged, and that's the development of an evidentiary

1 base for these various technologies.

2 MS. BERRY: Muin?

3 DR. KHOURY: I think this is where the rubber hits the road. We have  
4 been talking about these issues now for about five to 10 years, dating back to Tony Holtzman's  
5 sort of NIH/DOE task force on genetic testing. I guess as a public health agency, and I'll  
6 present some of this more tomorrow morning, we have been looking for ways to supplement the  
7 oversight mechanisms that FDA and CLIA have in terms of assuring the quality of services and  
8 using evidentiary-based models like U.S. Preventive Services Task Force.

9 We have played around with different kind of model processes that I'll  
10 present tomorrow morning, but I think there may be another option for developing the sort of  
11 evidence-based approach as we move forward. Let me give you an example. We have been  
12 working very closely recently with the U.S. Preventive Services Task Force and AHRQ using  
13 the experiment of BRCA1 testing as an example, using one of their evidence-based centers, and  
14 looking at the evidence.

15 We have been meeting with various folks about this. It is very obvious that  
16 for most of genetics testing and technologies, what the U.S. Preventive Services Task Force  
17 would do is return insufficient evidence as their criteria. So this is a problem, because in the  
18 meantime, these tests are widely used. There are obviously  
19 direct-to-consumer campaigns about them, and so what we need to do is develop a process that  
20 can guide and help bodies like the U.S. Preventive Services Task Force, and other technology  
21 assessment processes, to do a first look at genetic technologies when they come out of the  
22 research oven, identify the gaps, and then use the processes of data collection that are a bit  
23 more targeted using public/private partnerships as models. Some of you may have heard me  
24 sing this tune now for five years --

25 DR. McCABE: Yes, we've been listening to this for 10 years, Muin.

26 DR. KHOURY: No, actually you're absolutely right. You've been listening  
27 to me for 10 years, so bear with me. I think there will be another process that I will present to  
28 you tomorrow, another experiment, a three-year experiment to see how we can do this.

29 But to me, that's the most important missing link between research and  
30 practice, is the sort of how do we implement an orderly process of transition that uses both the  
31 regulatory oversight mechanisms that already exist, along with processes of public and private  
32 partners coming together to weigh in on what is ready for prime time, and what is not ready for  
33 prime time. I think I'll just end here.

34 MS. BERRY: I have a suggestion. I don't know if this lends itself to it or  
35 not, but I sort of like the process we went through in terms of prioritizing our larger issues. I  
36 wanted to throw this out there for the group, because there are so many on this list here of  
37 possible topics for a recommendation, whether we should undertake a similar effort here where  
38 we determine what should be the focus of our efforts, and kind of prioritize amongst ourselves  
39 and see what that comes up.

40 It is really a daunting list. Now, others may say you know what? There is  
41 no reason why we can't look at each and every one of those issues and develop  
42 recommendations for each. But I suspect that that wouldn't be quite as helpful to the Secretary  
43 or anyone else if we were to cast that broad a net, and perhaps we should be a little bit more  
44 focused on some of the recommendations that have the greatest likelihood of producing an  
45 impact.

1 I throw that out for discussion, but I'm trying to figure out how do we move  
2 from kind of the theoretical, we kind of know what our goal is, but to get to the nitty gritty of  
3 where should we focus our deliberations.

4 DR. McCABE: I think that is probably a very good idea, because I think  
5 with this list, nobody can keep in mind a list this long, let alone the Secretary and his office,  
6 with all the things that he has to deal with. So I think that would be a very good idea.

7 MS. BERRY: Yes, Sherrie?

8 DR. HANS: I also think that is a terrific idea. I just wondered whether we  
9 wanted to sort of finish with any general comments first before moving onto that. I had just a  
10 couple of ones I wanted to get on the floor, and I don't know if there were others.

11 Just really quickly, and the reason I wanted to sort of make them publicly is  
12 just so people could think about them, particularly during the recommendations. I have more  
13 detailed written comments that I can give to the staff. One is to just pick up on an earlier  
14 theme.

15 I think that Toby Citrin in particular framed well, staff did a fabulous job of  
16 putting this document together and covering a very broad area. The one area where I would  
17 encourage them to work a little bit more on is making a stronger statement of need. In  
18 particular, framing out some of the cost issues.

19 Why is this something important to consider now? What kinds of dollars  
20 are we talking about? How quickly is this an emerging issue? If I'm the Secretary, why should  
21 I care about this today? I thought that there needed to be more of that in the front material in  
22 the report.

23 The other piece, which may come up in the specific recommendations, and I  
24 apologize that I wasn't at the previous meeting, and perhaps this was covered at that time. I was  
25 a little bit concerned about some of the statements about the sizable portion of costs going  
26 toward royalty fees. I wasn't sure whether there was actual documentation such as a survey that  
27 documented that as a statement, and I don't know whether that was simply through testimony. I  
28 just wanted to sort of put that marker in there that if it is supported by really good evidence,  
29 that should be included in the report and be considered for folks making recommendations. If it  
30 is, then it needs to be noted in more couched language, that it was brought up in testimony and  
31 through comment, but I'm not sure that we have a handle on exactly how big that problem is.

32 MS. BERRY: Did staff want to comment?

33 MS. GOODWIN: The evidence provided in the report was primarily given  
34 by Andrea Ferreira-Gonzalez at our last meeting. And actually Debra might be able to  
35 comment. Mildred Cho has published a few papers, and I think it goes into this topic a little  
36 bit. Is that right?

37 DR. LEONARD: Well, Mildred Cho, John Mertz, and I have worked  
38 mostly on the availability of testing, and the impact on gene patents. I don't know that the  
39 question has directly been addressed as to the cost. But it certainly has had a significant impact  
40 on availability of testing, because 25 to 50 percent of laboratory directors report that they have  
41 not done, or have had to stop doing testing, because of gene patent enforcement.

42 That is shutting down laboratories, but then when you can license patented  
43 technologies, the costs of those are exorbitant, relative to what we get paid for doing the  
44 testing. That is data that is hard to collect, because usually there is a confidentiality, or like a  
45 keep your mouth shut about what the conditions of the license are, so that most people who

1 have a license can't talk about how much it costs them, and what the royalty fees are, because  
2 part of the agreement is that you can't say.

3           So that would be information that is very hard to collect. But the up-front  
4 licensing fees are anywhere from \$10,000 to over \$100,000, and then a per test fee of anywhere  
5 from I think I've heard up to \$60 per test, for a range of different genetic patents.

6           DR. HANS: And I would just encourage the staff to put in more nuance  
7 statements that capture some of what we just heard here, that isn't sort of a definitive statement  
8 of you can point to a particular study or collection of information.

9           DR. LEONARD: But hopefully part of this data could be collected through  
10 the NAS study that is being done on gene patent's impact on research and health care services.  
11 But I don't know if they're going to collect that information.

12           MS. BERRY: Debra, did you have anything additional beyond responding  
13 to that? I had written down that you had --

14           DR. LEONARD: Well, I was going to suggest that staff has come up with  
15 various policy options, and maybe one way to do this is to go through the policy options, and  
16 then go back and see if there are other areas where policy options have not been recommended  
17 where we may be able to address some of the issues. But that may be a way of working  
18 through the various issues that we have to address.

19           MS. BERRY: Joan?

20           DR. REEDE: I just had a question in terms of this issue around the  
21 royalties and reimbursement. Is there anything, or would it be useful to have something that  
22 required transparency in terms of that information for reimbursement to occur?

23           DR. LEONARD: It would be great if there was a CPT code that could  
24 capture that, but the problem is that then a reimbursement level would be set for that CPT code  
25 that probably would not reflect the actual cost of the royalties, because they would vary.

26           A number of discussions have looked at ways of having like mass  
27 bargaining for different gene patents so that it wasn't laboratory by laboratory, but there was  
28 more standard policy on how much is a reasonable amount to charge per test for royalty  
29 payments. I'm not aware of anything moving forward on that front, but it would be great if  
30 there could be transparency.

31           The problem is that all of the licenses that I have seen, and contracts,  
32 specify that you cannot discuss any of the terms of the license agreement once you sign. So I  
33 don't know how you would achieve that transparency. Maybe a lawyer in the group could say  
34 whether those two things are contradictory, I don't know, but it would seem to me that they are.  
35 But for health care coverage, it would be very nice to have those out in the open, so more  
36 people were aware of what they were paying for, and what the cost of gene patenting and  
37 enforcement is on health care costs.

38           MS. BERRY: Emily?

39           DR. WINN-DEEN: So I had a couple of things that I thought were  
40 important for us to talk about that aren't on the topics list. They were actually addressed in the  
41 body of the text, but somehow didn't make it onto the pullout list. One is time dilemma in  
42 terms of real preventive medicine where the cost of the test would be borne probably by  
43 younger people on private insurance, and many of the benefits would be reaped by older people  
44 on Medicare.

45           So you have different parts of the payer organization bearing the costs and

1 getting the benefits. So this deals with sort of common complex disease, so when you get into  
2 cardiovascular preventive medicine, diabetes preventive medicine, and those kind of diseases,  
3 we get away from monogenic kind of disease.

4 I think that is something that in the U.S. system, we are going to have to  
5 grapple with. The systems that have nationalized health care don't have to worry about that,  
6 because they very clearly see the benefit.

7 The other thing is we have evidence-based coverage decisions, but we  
8 haven't really talked about the health economics of that. So in terms of making a case for  
9 preventive medicine, do we need to just make a medical case? Do we need to make a health  
10 economic case? And are we only going to get coverage if we make a health economic case, and  
11 not just a medical case?

12 So I think that is all part of that whole preventive medicine shift from  
13 thinking about genetics just in terms of pediatric, neonatal screening, monogenic disease, and  
14 getting into sort of the next wave of the genetic component of most major diseases. And  
15 somehow we have to deal with that, or at least highlight it as an issue that needs to be dealt  
16 with in the future, if not immediately.

17 MS. BERRY: How about the threshold issue? It is a threshold issue to me,  
18 but maybe it is clear to everybody else. Do we need, or want, to be specific as to what types of  
19 technologies we're talking about?

20 We use these terms very loosely, genetic technologies, genetic tests and  
21 screenings, and I guess this gets to the evidence-based discussion. Are we at the point where  
22 we can establish guidelines or guidance, or help the Secretary come up with, and this gets to  
23 things that Reed was talking about and others, ways to assess whether something has value and  
24 should be incorporated in the whole health care diagnosis and treatment system? I don't know.  
25 Do we need to go down that path, or do we purposely leave it vague? We just keep talking in  
26 very general terms, but we all know from previous discussions that some tests are better than  
27 others, and some have validity, and some don't. Or has that bridge already been crossed, and  
28 we don't need to worry about that?

29 DR. TUCKSON: This is Reed. I would hope that we would be able to  
30 start, at least for our discussion of these issues, to be specific about examples. For example, if  
31 you take the DNA tool assay exams, those are very professionally checked. They have a  
32 meaning for what it will or will not be in terms of diagnostic colonoscopies going forward.

33 It has something to do with whether or not we will (inaudible) regular fecal  
34 occult blood. So that is a very concrete example of a kind of test. As we have alluded to, there  
35 are things that have to do with is your kid going to have blue eyes sort of thing, which is a  
36 different kind of category.

37 I think that it would be useful for us to have some specific categories of  
38 these things as we try to dig a little deeper into (inaudible) as finding examples that highlight  
39 and illustrate the real policy interventions that we're trying to concern ourselves with.

40 MS. BERRY: Debra?

41 DR. LEONARD: Well, in reading this, I went back to SACGT's definition  
42 of genetic test. In reading this, particularly in Policy Option 1, a lot of it is based on family  
43 history, and focuses on truly inheritable germline variations or mutations that cause disease  
44 risk.

45 The SACGT definition was much broader, and went beyond true germline

1 genetic tests, and included somatic mutations, and potentially even infectious disease  
2 applications. And so how those are treated is very different than those genetic tests and  
3 services that are related to inheritable germline variations and mutations that correlate with  
4 disease risk.

5 I don't know if we need to go back and revisit the SACGT definition of  
6 what genetic testing or genetic services were, but it still remains. That was one point that was  
7 very controversial at the time that SACGT developed that definition.

8 MS. GOODWIN: Can I actually draw your attention, Debra, to page 13 and  
9 14, where we propose the definition for genetic technologies and genetic services to kind of  
10 frame the discussion in this report, and certainly taken from that SACGT definition, it is altered  
11 a bit to better reflect the different discussions. So rather than taking the definition from a more  
12 oversight perspective, we are trying to broaden it for the purposes of this topic.

13 DR. LEONARD: But then when you put such an emphasis on family  
14 history, somatic mutations are not included in family history, unless they are genetic cancer  
15 syndromes. And so I'm concerned about putting somatic mutations into the same category as  
16 germline mutations and variations, since you will not pick up somatic changes by family  
17 history.

18 DR. WINN-DEEN: Yes, I just want to concur with Debra, that I think we  
19 have to be really clear when we're talking about cancer genetics, about whether we're talking  
20 about cancer syndromes, or tumor profiling. If we're going to expand genetic testing to include  
21 tumor profiling, which has been suggested, and a lot of the --

22 DR. LEONARD: Well, it is part of the definition.

23 DR. WINN-DEEN: Right, right.

24 DR. LEONARD: The existing SACGT one.

25 DR. WINN-DEEN: Right. But we just need to be very clear about when  
26 we're talking about these things, which scenario we're talking about, either somatic genetics, or  
27 germline genetics.

28 MS. BERRY: Muin?

29 DR. KHOURY: Actually, if we take colorectal cancer as an example, you  
30 can have the whole spectrum or continuum of genetic applications, from the rare to the  
31 common. We need to be very careful, because at the end of the day, what we're trying to do  
32 here is figure out the value added of either a genetic test on the blood to determine germ  
33 mutations, or a genetic test on the stool to figure out whether there are DNA signs of the  
34 cancer, or a simple family history, too, that you administer and you find out that the person has  
35 a first-degree relative with early onset colorectal cancer.

36 We need to be able to use whatever SACGT has done, because they spent a  
37 long, long time figuring out the definition of genetic and genomic tests. And second, the  
38 process of validation of genetic tests. I mean, they use analytic validity, clinical validity,  
39 clinical utility, and the ELSI issues in a very detailed fashion, and they kind of laid it out like  
40 cold supper for us to implement.

41 So here we are talking about reimbursement of services, so you have to  
42 come back to what is it we are being reimbursed for? Is it a somatics test on this tool, or a  
43 genetic test combined with a family history, pedigree taken to find the people with HNPCC, or  
44 the APC syndromes for colorectal cancer, or somebody who has just a  
45 first-degree relative with colorectal cancer at the age of 65, and therefore, may need a



1 colonoscopy earlier than the general recommendation, which is after age 50.

2 So the bottom line in all of this is what is the value added? I think the  
3 health economics discussion is very apropos here, because in a time of limited resources where  
4 you are comparing options A and B, you have to kind of compare a genetic-based practice of  
5 medicine with a general practice of medicine that doesn't use genetics, and do a decision  
6 analysis, like they did with the TMP example in 6MP with treatment of acute leukemia that  
7 David Veenstra represented to you last time. So what are the pros and cons? And then at the  
8 end of the day, how much money are we going to spend if we use a family history/genetic test  
9 tool on some people, and a DNA-based assay on other people?

10 So the main recommendation here from me is take this high level document,  
11 and then begin to apply it to very specific case studies, perhaps guided by what SACGT has  
12 done for us over the last three years, because they lay out very clearly the types of genetic tests  
13 for diagnosis, prevention, prediction, and then the process of validation on how to do this.

14 DR. TUCKSON: Could I ask Muin to take it one step further? I wonder  
15 that after we've got all this analysis, what do you see as the level of detail coming out of our  
16 committee after we have done that? I think that is what I was trying to get at.

17 The question is, obviously we're not going to see ourselves then  
18 recommending a specific test for the Secretary to get access to, but this is leading to some sort  
19 of an algorithm for the next step of policy or process development in the Secretary's office.  
20 What do you see?

21 DR. KHOURY: I think, Reed, you should be able to determine this as a  
22 committee, because I don't see you getting into the real specific test by test discussion, but more  
23 of a high level guiding principle for HHS on how to go about doing this. You could be guided  
24 by the case examples as you go through the deliberations, but I see kind of an intermediate  
25 menu or cookbook in a way, but not very detailed, because that has to be worked out by the  
26 agencies themselves, CMS, and what they cover for.

27 But moreover, general guidelines, perhaps of the kinds that SACGT started  
28 doing during the tenure, and then leave it for implementation. But you can monitor the  
29 progress of HHS agencies.

30 DR. McCABE: I wanted to go back to the point of definition, and bring  
31 people back to page 13, where I think the implication, with a sidebar from staff, is that it was  
32 their intent that these be germline mutations here. It certainly says somatic genotypes, but if  
33 you look at all the examples, they are all germline.

34 DR. LEONARD: So can that somatic be taken out?

35 DR. McCABE: That's the purpose of this discussion, would be to get the  
36 feeling of the committee. We need to move forward on this, as Muin says. We have been  
37 debating this issue of definition also for 10 years.  
38 If you look at the examples, they are all clearly germline.

39 DR. LEONARD: In a way, this is kind of like the genetic exceptionalism  
40 issue. Sometimes it is good to lump it in, and sometimes it is good to exclude it. It is the same  
41 sort of thing with somatic mutations. When you are talking about genomics, medical advances,  
42 and improving health care, it is probably good to include the somatic mutations.

43 But when you are talking about the kind of regulatory issues of appropriate  
44 counseling, appropriate ordering of the tests, and getting the testing through family history,  
45 those only apply to germline mutations. So I guess there are times when you would want the

1 definition to include all types of genomics beyond just inheritable, and there are other times  
2 when you really need to distinguish between somatic and germline, because the implications  
3 are very different.

4 Somatic mutations have no implications for the rest of the family members,  
5 whereas germline mutations, you are dealing with an entire family.

6 DR. McCABE: And so that if we took it out of the definition in the sidebar  
7 here, since in fact all of the rest of that sidebar relates to inherited, then you could put some sort  
8 of a thing at the end. The title here is "What are Genetic Technologies?" as opposed to  
9 genomic technologies. Either in the text or somewhere else, you could put that similar  
10 approaches may be used for looking for somatic mutations using genomic technologies.

11 DR. LEONARD: And even infectious disease, because part of our charge is  
12 bioterrorism, although we don't look at that that much, but infectious disease and emerging  
13 infectious organisms, that type of thing also benefits from genomics, but they don't have the  
14 same familial component.

15 DR. McCABE: I guess it is a judgment that you all have to make, what to  
16 include or not include. I'm just reacting to what Ed said about similar approaches can be  
17 developed for genomic tests.

18 I think what comes to my mind immediately is that those approaches are not  
19 similar. I mean, what applies to germ cell is different from what applies to somatic cells. I  
20 think we are going to be faced increasingly with the mixing and matching of both technologies.

21 So if you have a first-degree relative with colorectal cancer, you may use a somatic cell to find  
22 a somatic diagnostic test on the stool to find early evidence of cancer, rather than colonoscopies  
23 on the proband.

24 So we are going to be mixing and matching somatic and germ cell  
25 mutations, so that's number one. Number two, even among germ cell allelic variation, we do  
26 have to clearly separate the high penetrance rare alleles, the PKU, the Huntingtons of the world  
27 type diseases from the day to day SNP variation that is associated with low disease risks and  
28 complex gene/gene and gene/environment interaction. Because those kinds of genes do not  
29 lead to significant familial aggregation, and their use as a technology is likely to be bundled up  
30 together in genomic profiles that have very different kind of a more sturdy, I guess, validation  
31 process, if you will, than the simple single-gene genetic disorders.

32 So as we go through this process for reimbursement and coverage of  
33 different tests, I guess you start by asking the high level questions. Are the tests analytically  
34 valid? Do they predict any clinical outcomes? More importantly, can they change the outcome  
35 in reducing morbidity and mortality? And what are the ethical implications?

36 And then as you go through this armed with the processes of different  
37 groups like the U.S. Preventive Services Task Force, each one of these types of tests may have  
38 a different threshold for crossing between what is ready for prime time, and what is not ready  
39 for prime time. We spent quite a bit of discussion in SACGT about rare diseases, because  
40 those rare diseases, and I guess this committee has taken it on too, may have a different kind of  
41 a threshold, because of the difficulty in collecting data because diseases are rare, and the  
42 validation process therefore may be either delayed, or non-existent to begin with.

43 So we have the whole gamut here of tests from somatic to germ cell, and  
44 within the germ cell you have the highly penetrant single-gene diseases to a bunch of SNPs,  
45 polymorphisms, and gene expression profiles that you use in future bundling, either for

1 pharmacogenomics, or just genomic profiles of the kinds that some companies are sending us  
2 right now prematurely for preventive medicine.

3 MS. BERRY: Ed, and then Agnes?

4 DR. McCABE: My point was purely practical. Having heard these  
5 discussions for the past decade and been a party to them for the previous three years,  
6 recognizing that the division between somatic and germline can get a little bit fuzzy where  
7 sometimes somatic is superimposed upon germline, as opposed to as far as we understand,  
8 purely somatic at this time, though there may be some predisposition. It was just a way of  
9 moving on, getting beyond that, and at least getting somewhere by limiting the definition to  
10 germline, and taking it out of the debate that has to do with these definitions that are used in the  
11 clinical laboratories.

12 So that was my purpose. I think we could say let's focus on germline for this  
13 document, let's recognize that the lessons we learned here may be applicable to somatic, but  
14 let's at least move forward at this point. That was the purpose of my comments.

15 MS. MASNY: I was thinking more along what you were saying, Dr.  
16 McCabe, that if you divide it that way, then I think we may be defeating the purpose of what we  
17 were trying to even do earlier looking at the education of the general health force. If you are  
18 only looking then at the germline mutations, then the focus on the genomics and where most of  
19 the actual practice in genomics is going to be done in the primary care, if we don't focus on that  
20 and address some of the reimbursement issues, we know that reimbursement very often drives  
21 practice, as well as what people implement.

22 So I think that we should continue, if we do that, we're looking specifically  
23 at the genetic technologies, and focus on the germline mutations. I think maybe if we had not  
24 just at the end of the document, but somewhere right after this, another sidebar looking at what  
25 are genomic technologies, so that they sort of are dealt hand in hand the way we did in our  
26 previous documents where we were putting the slash between genetics and then genomics.

27 I think it has to be addressed. I do think even from the level of somatic  
28 mutations, it will involve family history. Because, for example, in the ovarian cancer testing  
29 that they are proposing, the use of proteomics, they are already talking about looking at  
30 guidelines for who would be best to use this test based on family history. So although the  
31 proteomic may be picking up as certain somatic patterns or tumor profiles of those protein  
32 patterns in an ovarian cancer, they still would be using family history to determine who would  
33 make use of that test.

34 DR. McCABE: Yes, I was just talking to Suzanne, and certainly we could  
35 add a sidebar that would deal with the somatic and other genomic technologies, even getting  
36 into some of the issues about genomic technologies applied to microbiological organisms. I  
37 understand that a lot of what we do are really the applicability to the general population, that  
38 are going to go beyond this. I think that it is important that we move forward after a decade,  
39 and not get hung up again here.

40 MS. BERRY: Debra?

41 DR. LEONARD: Well, in the context of coverage and reimbursement, the  
42 genetic counseling only applies really to the germline mutations. When you talk about the  
43 testing services, laboratory tests are all coded with the same CPT codes, regardless of whether  
44 you are talking about germline, looking for germline mutations, or looking for somatic  
45 mutations.

1                   So in the context of coverage and reimbursement, it is all the same codes  
2 anyway, regardless of whether you're doing somatic or germline, or whatever, and they are all  
3 inadequate.

4                   DR. McCABE: So by including them as sidebars within here, but  
5 separating them out, perhaps we could try to make those distinctions, and not have them  
6 continue to be blurred.

7                   MS. BERRY: It seems to me that that is really a threshold question. What  
8 are we talking about when we're talking about coverage and reimbursements? Coverage and  
9 reimbursement of what? And then we seem to go and we take a leap, and we all do in our  
10 discussions, the report, and public comments. Genetic technologies need to be covered and  
11 reimbursed.

12                   We have to back up a little bit, and this gets to Reed's point and Muin's  
13 point, and others, we are not really saying that. We are not saying that all technologies need to  
14 be covered. So perhaps there is something that we need to do in the way of recommendations  
15 pertaining to what scientific evidence is necessary, where are the gaps that would help  
16 decisionmakers, be they federal government programs, be they private insurers, make the right  
17 decision in terms of coverage, and subsequently, reimbursement.

18                   DR. WINN-DEEN: So I think one of the things that is important to think  
19 about when you come to predisposition or risk assessment testing, is in much the way that  
20 family history has been used in the past, this testing is quite likely to be used as some kind of a  
21 gatekeeper for more intensive monitoring.

22                   So either you start earlier, you do it more frequently, and to Agnes' point,  
23 you identify people who would benefit from expensive testing, or you never would recommend  
24 it for general population screening, but for a subset of people who are "at risk."

25                   And so I think we need to think about also the trickle down effect of a risk  
26 assessment test as a gatekeeper for other more traditional monitoring tests, you know, BRCA1-  
27 positive individuals start mammograms earlier, or start mammograms a whole lot earlier than  
28 general population screening.

29                   There are good monitoring things. Not only do we need to worry about  
30 whether the primary gatekeeper test is covered, but also once people are put into a risk category  
31 based on that gatekeeper test, is their monitoring also covered? Because that then would be  
32 they are not symptomatic, so it is still presymptomatic testing, but it is absolutely medically  
33 indicated, or we all believe it is, anyway. So I think that is another wrinkle that we somehow  
34 have to consider at sort of the risk assessment test as a gatekeeper for more standard monitoring  
35 analysis.

36                   DR. LEONARD: And not only the more expensive monitoring, but also the  
37 follow up things that may be done, like prophylactic mastectomies. Will they be covered? The  
38 real sort of treatment choices that people who are identified then as being at high risk may  
39 choose to have done.

40                   DR. WINN-DEEN: Yes. So the whole preventive medicine strategy,  
41 whether it is a colostomy, a mastectomy, an oophorectomy, whatever.

42                   DR. TUCKSON: This is Reed. I tried to write a lot of my comments down,  
43 and I think you all have copies of it, so I won't waste your time rehearsing any of that. But  
44 what I think I like about what just got stated is it is that level of detail that I think we need to,  
45 and I think this may be what Muin was getting at a little earlier, is that as we start to work some

1 of those out, we then by delving in that kind of depth of the downstream consequences of some  
2 of these things, that we start then to think about what our policy recommendations might need  
3 to look like.

4 What I'm sort of getting at here is that things have consequences. One of  
5 the things I hope is that we don't wind up putting ourselves in a position wherein advocating for  
6 a good thing, that it means that automatically a new test, because it is available and exciting and  
7 good, becomes the subject of something that is automatically reimbursable, when it may result  
8 in five other things needing to occur that are associated with it.

9 Maybe those things are appropriate, maybe not. But then that one new thing  
10 causes maybe four other diagnostic restudies to confirm or whatever, and then the economics of  
11 that get to the point where it becomes crazy. At some point in this, I just want to make sure that  
12 we don't position ourselves as being irresponsible to the notion that things occur in context to  
13 other things, and how do you think about that in a world of real constraints is, I think,  
14 something that we could add. How do you approach those kinds of issues, or do you simply say  
15 let whatever happens, happen, and that is not our responsibility to think about.

16 MS. BERRY: Ed?

17 DR. McCABE: You know, I think it would be worthwhile to look at the  
18 recommendations now to give staff some assistance with how to couch those. And Cindy  
19 before said maybe we should prioritize those. I went through and began to categorize them, but  
20 I should have looked over my shoulder and seen that staff had done a much better job than I did  
21 on the fly here.

22 So that if you look at the PowerPoint slide there, it really falls into four  
23 categories. Even if we take the broader issues and separate those out, we're down to five  
24 categories. So that it is a way to organize by Medicare, and what the issues are there for  
25 Medicaid/SCHIP, then what the issues are for all insurers, and then the broader issue.

26 So what we might do is look at how we could prioritize within each of those  
27 categories perhaps if we feel the need to do that, but at least it is an organizing principle that  
28 allows us to go from whatever the number was before, down to four or five categories. So  
29 perhaps there could be some discussion within each of these categories now.

30 MS. BERRY: Emily?

31 DR. WINN-DEEN: So can I just ask sort of a point of order? Because if I  
32 had to look at those and say, should we talk about all insurers, or just Medicare, because all  
33 insurers encompasses Medicare and includes a whole lot more people, I would say, do that.  
34 But in terms of what HHS is directly responsible for, it is only Medicare.

35 So could you just give us a little committee guidance? Should we focus on  
36 issues that HHS can control, rather than the whole global insurance industry? Or how do we do  
37 that?

38 DR. McCABE: I certainly think we could comment on all, but in terms of  
39 what the Secretary has control over, I would think it would be those things that the Secretary  
40 has control over, so that we could perhaps have some guidance from the ex officios with  
41 respect to what they think, where they think this committee could have the greatest impact. It is  
42 Medicare and Medicaid, and also then the other agencies sitting around this table. So could we  
43 have some help from the ex officios, perhaps, with respect to where you see the biggest bang  
44 for this committee's efforts?

45 MS. BERRY: And if I can just sort of add to that. Are there studies, either

1 going on right now, or possibly being contemplated or studied, that perhaps we could suggest  
2 that the Secretary commission either through one of the agencies, or from the outside, I don't  
3 know the most appropriate way to do it, that would get to this issue of evidence-based  
4 decisionmaking that could be while not directly influencing private health plans, certainly if  
5 there were a government-sponsored study, that could influence private health plans indirectly,  
6 in addition to informing Medicare program. I don't know the answer to that.

7 DR. KHOURY: Is Reed still here?

8 DR. TUCKSON: Yes, I'm here.

9 DR. KHOURY: I guess this committee can make an influence in a number  
10 of areas. Obviously HHS direct jurisdiction is Medicare/Medicaid. But HHS presides over  
11 NIH, CDC, FDA, and AHRQ. As you said, Reed, earlier, that recommendations from the U.S.  
12 Preventive Services Task Force count a lot with respect to the private world, isn't that correct?

13 DR. TUCKSON: Yes, very much.

14 DR. KHOURY: So basically what you are recommending to the Secretary  
15 is not only something that would influence Medicare and Medicaid, but sort of this convening  
16 role that hopefully by doing other things, like evidence-based coverage decision, will influence  
17 the whole practice of health, health care, and preventive medicine in this country for the private  
18 sector as well.

19 So I don't feel like you need to be constrained necessarily by this hierarchy  
20 of Medicare/Medicaid, nor do I think that a recommendation should be different for different  
21 groups, because what you want to do is lay down the basic principles and guidelines for what  
22 should be covered and not covered, and reimbursed or not reimbursed, and then it will play out  
23 through the various processes of the public and private sector.

24 I mean, you are tackling here issues that are relevant to the practice of  
25 medicine and health in this country that are way beyond genetics. I don't think we're trying to  
26 fix medicine here through the lens of genetics, but to try to see how those new technologies fit  
27 in the underlying scheme of the practice of health care in this country. But that is my opinion.

28 MS. BERRY: Yes, Kaytura?

29 DR. FELIX-AARON: I agree with you. I mean, I am struggling, because  
30 I'm not sure that I understood the question that you posed, but I just wanted to respond to  
31 Muin's point. The U.S. Preventive Services Task Force addresses really basic questions. Is  
32 there enough evidence to recommend a particular service? You get a number of letters.

33 The challenge here is that the evidence, and I go back to Steve's point about  
34 the evidence base for making those types of evaluations is that chances are that evidence base is  
35 really often non-existent, and so the recommendation that comes out is I, insufficient evidence.  
36 Not that it is discouraging those services, but that there is not sufficient evidence.

37 So I think that the U.S. Preventive Services Task Force does a lot of good  
38 work, but I haven't heard around the table sort of questions that we have that would be  
39 appropriate for that particular body. However, AHRQ also has the evidence-based practice  
40 centers, which addresses more policy, sort of like higher level questions. If this committee had  
41 a particular question that it wanted to address, our EPC programs could address a particular  
42 question.

43 The EPC program is such that it not only relies on the published literature,  
44 but is increasingly looking at the gray literature, literature in reports, and increasingly pooling  
45 reports and experiences, sort of practical experiences to draw conclusions, and to provide

1 guidance.

2 So provided that this group came up with a clear question and wanted  
3 guidance on how it ought to tackle a particular question that had policy relevance as well as  
4 clinical relevance, the EPC program and AHRQ could address that.

5 DR. McCABE: I've asked Cindy's permission to come to the mike at the  
6 podium, just to save my neck, trying to crane 180 degrees. I think if you look here, let's look at  
7 the recommendations, the possible topics for recommendations.

8 As I look through the all insurers category, I don't really see any topics there  
9 that don't apply to Medicare and Medicaid, and I see some that actually collapse into some of  
10 the other topics. So that if we look, for example, for coverage decisions, under that, there are  
11 issues like the screening exclusion under coverage decisions. Also, if we go under all insurers,  
12 we see evidence-based coverage decision, we see a need for a, rather than a lack of uniformity,  
13 we could say a need for uniformity to make it more positive in coverage decisionmaking. Also  
14 reimbursement determinations come to some extent, under that.

15 Basically all of these issues apply to Medicare and Medicaid, as well as to  
16 all insurers. I mean, that's by definition, Medicare and Medicaid are all insurers. The issue is  
17 is there anything there that we should exclude as being too numerous in terms of the menu to  
18 the Secretary? Or should we look at collapsing them into fewer categories as a way of  
19 organizing this.

20 MS. BERRY: Steve?

21 DR. PHURROUGH: Well, I'll try and give you some concrete  
22 recommendations. I think the issues as you were, I think, trying to elaborate, Ed, are not  
23 around the kind of payer. The issues are around what is the evidence base, what are the barriers  
24 to applying the technology using that evidence base, and then perhaps there are some specific  
25 Medicare or alt payer issues.

26 So I would say that the first priority is developing an evidence base, and it is  
27 a recommendation that the Secretary task his appropriate agencies, which I think are AHRQ,  
28 NIH, CDC, and perhaps even HRSA, with systems research to commission a technology  
29 assessment that defines the current genetic technologies, the evidence base for those, and  
30 identifies the gaps and knowledge it needs further trials to determine. I think that's number one.

31 I think number two is define the Secretary's task CMS, and in fact we've  
32 already made our recommendation that the Secretary provide in its recommendations to  
33 Congress, whatever that is, that the screening evaluations become a routine part of the  
34 Medicare portfolio, and that CMS be allowed to make those decisions based upon the best data  
35 that is at hand.

36 Three are other barriers, such as CPT codes and reimbursement  
37 determinations, and we could ask the Secretary to task the appropriate agencies to define, well,  
38 I'm running out of concrete ideas. I think rather than saying what should Medicare do, and  
39 what should all insurers do, I think there are things across the realm, and I think the first is the  
40 evidence base, and I think the other is incorporating screening into the entire insurer  
41 population, and not just private.

42 DR. McCABE: But using evidence base in the grandeur, not U.S.  
43 Preventive Services Task Force's slow, methodical, perhaps too slow and too methodical  
44 sometimes approach, but look at where utilizing best practices and literature could define an  
45 evidence base for this, the way we use it more typically in medical practice.

1 DR. PHURROUGH: Exactly.

2 DR. TUCKSON: I've got to sign off. Can I just make one quick comment?

3 DR. McCABE: Of course.

4 DR. TUCKSON: Thanks, Ed.

5 I like what we just heard from AHRQ, I think it sounds good, and I think we  
6 ought to fall for those kinds of larger institute studies that AHRQ can do outside of the U.S.  
7 Preventive Services Task Force.

8 The only other comment that I would want to emphasize though, is that  
9 there ought to be very clear criteria for when recommendations about going forward for  
10 extensive things ought to be done when there is no evidence basis. I can clearly understand  
11 there are times when you have to go forward, but I think we've got to be very clear about what  
12 may precipitate that recommendation.

13 Secondly, in my analysis, I did call for a little attention on looking at the  
14 actual quality of performances as based upon professional guidelines for the delivery in this  
15 area. The guidelines need the big step from the evidence, and then evaluation of what you  
16 actually paid for from the taxpayer's point of view, and private purchasers based on those  
17 guidelines. That is something that we didn't get into, and I just want to put it on the list for  
18 consideration maybe sometime going forward. Thank you very much.

19 DR. McCABE: Thank you for being with us this afternoon, Reed.

20 DR. TUCKSON: Thanks.

21 MS. BERRY: Emily?

22 DR. WINN-DEEN: I guess I just want to second that comment that maybe  
23 the broad categories are one, coverage. What is needed to establish that a test should be  
24 covered? This goes back to SACGT's long discussion about clinical utility, when is clinical  
25 utility established, and then what are the other things that follow from that?

26 Then the second thing is once you have determined that something should  
27 be covered, how do you assure adequate reimbursement, and that gets into this whole is the  
28 CPT code system working, is the reimbursement associated with method codes the right way to  
29 do it? That's another whole set of discussion items.

30 Then I like the sort of final everything else broader issues category of things  
31 that if coverage was there and reimbursement was there, what are the other things that we  
32 would need to be concerned about in bringing this to the practice of medicine? So what are the  
33 remaining barriers to entry? Because I think a lot of people think that they are all coverage and  
34 reimbursement, and I think we clearly in our morning session on education and training,  
35 identified that if a doc doesn't know how to use something, or a system doesn't know how to  
36 provide the right context for it and make people aware that they even should be tested, that it  
37 won't be utilized either.

38 I think if we divide things into those set of bins, it would help our  
39 discussion, and help make very focused recommendations. How do we improve coverage?  
40 How do we improve reimbursement? What are the things within HHS that can be activated in  
41 each of those areas? It just might simplify our job.

42 DR. McCABE: May I just ask one very specific question? So would  
43 people agree that one of the things that the Secretary might do would be to request input by  
44 whatever mechanism, specific mechanisms, have been recommended, but request input on how  
45 to improve those coverage decisions? Because that is the kind of thing that we can



1 recommend, that a study be chartered, or whatever.

2 DR. WINN-DEEN: Right, and I think just one little sidebar, there is an  
3 NCCLS group working on setting a guidance document on determining clinical utility. For  
4 example, I think it would be worthwhile for us to look at that draft document at the point when  
5 NCCLS is ready to share it, to see if that kind of a consensus process, which is what NCCLS  
6 goes through, would also serve us, separate from some government processes that might also be  
7 in the works to set out what is evidence-based medicine, and how do we know when we've  
8 gotten there?

9 MS. BERRY: Kay?

10 DR. FELIX-AARON: I wanted to draw the committee's attention to the fact  
11 that groups in the private sector, our federal partners, often request that the agency conduct  
12 studies. For example, the AMA could come to us and say, I want you to conduct a study on  
13 health literacy, and we do those studies.

14 So I wanted to raise again, it could go through the Secretary, and Sarah, I  
15 think you would have to guide us in terms of what types of interactions this committee can have  
16 with the agency. But in addition to going through the Secretary, it may be possible for this  
17 committee to actually request the agency conduct such a study, and that this committee may be  
18 able to go directly to the agency and work with the agency to create, implement, and conduct a  
19 study that addresses the specific questions we have laid out this afternoon.

20 MS. BERRY: Debra?

21 DR. LEONARD: I guess I have kind of a generic question. We talk about  
22 developing an evidence base, but really that is done one disease, one test at a time. I don't see  
23 how you develop a process to do that, because those who would need to be at the table would  
24 differ, depending upon what disease or what test you're talking about as to whether there is  
25 clinical utility for doing that. So can you create a generic process that would work for  
26 everything, for making coverage decisions?

27 DR. McCABE: After the experience with the SACGT, I would argue you  
28 probably could not make a generic decision that would cover everything, but that should not  
29 prevent us from getting started somewhere. So you can focus on what it is that you can begin  
30 to make decisions about, and identify a process for that, at least.

31 DR. PHURROUGH: Well, there are fairly standard processes that most  
32 payers go through in making coverage decisions. They vary somewhat in process, but the  
33 concepts are fairly similar. What we would like to see, those of us who are making these  
34 decisions, are to begin with, is sort of what is the state of the science now?

35 The state of the science isn't gathering experts together. The state of the  
36 science is let's pool through our normal processes, every piece of literature that has been  
37 published, and determine based upon that, what the state of the art is today. And then there will  
38 be gaps, and you'll list all the tests that are there, and you'll see where the gaps are. You'll list  
39 particular tests that have been proposed, perhaps they may have utility, and there will be  
40 complete gaps, because there is no published literature there. But evidence-based medicine by  
41 most carriers, by most payers, is an expert opinion. It is what has been published in the  
42 literature, based upon clinical trials.

43 DR. LEONARD: Can I follow up? So then the question is if you go  
44 through this process and identify the gaps, it seems that there are tests out there that are being  
45 used, but they are not covered, and there may be clinical utility, but it is not in the published

1 literature. Could then a request be made to NIH to put out an RFA for this series of diseases or  
2 tests for there to be funding to develop the evidence base, so that there could be coverage?

3 DR. PHURROUGH: I think the Secretary would be very hesitant to make a  
4 recommendation like that until we did have that evidence base.

5 DR. LEONARD: No, this is funding to develop, to see whether it should or  
6 shouldn't be covered. That requires research, and generally research that is not very well  
7 funded at this point.

8 DR. PHURROUGH: Correct. He would first want to know what is the  
9 state of evidence today, and then look forward, yes.

10 DR. LEONARD: Exactly. So there would be a body that would say okay,  
11 we've looked at these various tests and diseases that are out there being used, and for these  
12 three, we find plenty of evidence, and we're going to cover them. For these ten, we don't find  
13 enough evidence. Could that then be reflected in an NIH-type/NHGRI-type of RFA to ask for  
14 studies to address this?

15 But then you would also have to have guidelines out there about what kind  
16 of studies, what kind of scientific evidence you would need, how the studies should be done  
17 prospective, whatever, in order to provide appropriate evidence, but those types of studies are  
18 not well funded.

19 DR. PHURROUGH: That's correct.

20 DR. LEONARD: And so you never get there.

21 DR. PHURROUGH: A good technology assessment will tell you what  
22 trials need to be done, and be fairly specific, these particular tasks need these kinds of trials.  
23 And then I would assume this committee would make the recommendation to the Secretary that  
24 he task his appropriate agencies to look at those particular trials. It may not be NIH, it might  
25 well be AHRQ, and in fact, it could be CMS who may in some instances, provide  
26 reimbursement for technologies that aren't proven in the context of trials. We do pay for  
27 clinical trials. I'm not sure health systems research would fall into that category, but the  
28 Secretary can task his agencies to do whatever.

29 But I don't think he would do that until we first had what is the state of the  
30 art today, and what are the gaps, and then let me go out and fill those gaps. I think we need to  
31 study technology assessment first.

32 DR. McCABE: And I think what Debra is arguing is that there are quite a  
33 few gaps, if we have really started to look there. Certainly it fits with what CDC is doing, and  
34 what many of the agencies around here are doing, because it is recognized that there are huge  
35 gaps here. It clearly fits into the roadmap. You can't do translation if there is nothing to  
36 translate from, or to, which are some of these gaps that you would have.

37 I just wanted to go back over this organization, and be sure, because what I  
38 want to leave here this afternoon with is some guidance to staff about how to move from the  
39 document we have. We've had a lot of written input and a lot of oral input this afternoon. But  
40 the key is the recommendations, and we need to organize those.

41 So Steve, you had given us sort of an overview of an organization. Emily  
42 has, I think, taken that another step, and I want to be sure that everybody is in agreement about  
43 these categories, and then I want to talk about the broader issues and be sure that everybody  
44 agrees that they even need to be there.

45 So the coverage issues. The coverage issues, that would come under your

1 first heading, would it not, Steve? Because part of the coverage issues are that evidence base.  
2 How to assure adequate reimbursement? I don't remember what your second issue was, but  
3 would that be acceptable as a major category?

4 DR. PHURROUGH: I'm not real thrilled with a lot of discussion around  
5 reimbursement, but then that's because we argue reimbursement very commonly in CMS. But I  
6 think there are issues around what prevents coverage, and reimbursement is one of those. It  
7 may have a better title than reimbursement that I'm not thinking of at the moment, but  
8 reimbursement is an issue around why things aren't covered, and evidence is one of them. CPT  
9 codes is another.

10 DR. McCABE: I think for the practitioner community, one of the things  
11 we've heard over and over is that reimbursement is a huge issue, because people aren't doing  
12 these things if they're not getting reimbursed for them.

13 DR. PHURROUGH: We hear that for every single thing that we pay for.  
14 No one is yet to say we pay too much for something, we have yet to hear that complaint.

15 DR. McCABE: The real problem in genetics is, as Debra pointed out  
16 before, all genetic testing is covered under one code.

17 DR. LEONARD: Fourteen codes, to be exact.

18 DR. McCABE: Well, a very limited number of codes end up having to be  
19 creatively bundled and rebundled and everything to get anything to move forward.

20 What was your second category, Steve?

21 DR. PHURROUGH: My second category were Medicare-specific issues  
22 that all insurers don't have, and that's the screening exclusion issue. That's a specific Medicare  
23 barrier that others don't have.

24 DR. LEONARD: However, other insurers I have heard, speak and say that  
25 they don't cover preventive strategies. And so there are other insurers that do that. Not all, but  
26 some.

27 DR. PHURROUGH: By choice, not by law.

28 DR. LEONARD: Right.

29 DR. PHURROUGH: That's the difficulty we have, is the specific law that  
30 permits that.

31 DR. McCABE: Well, I need some guidance from the committee, then, so  
32 that we can leave here and have some instructions to staff.

33 DR. LEONARD: Can I comment? I don't care whether CMS likes the  
34 reimbursement issue or not. I think that this is a major issue, that the reimbursement is so  
35 inadequate, it doesn't even cover the cost. I'm not talking about the charge, I'm talking about  
36 the cost of doing the testing, so that it is a barrier to wide availability of the testing competition  
37 for pricing, and all sorts of other things.

38 If we are moving to genetic/genomic medicine in the future, that is going to  
39 be based on genetic/genomic testing that is not going to be paid for adequately to cover the  
40 costs. So I don't know, I mean, it is a very complex system of how reimbursement levels for  
41 CPT codes is set. I don't understand it completely, but it is not simple.

42 It is also constrained by the new Medicare Act that says that the laboratory  
43 fee schedule is frozen until 2009, and there are a lot of constraints on even addressing the  
44 reimbursement issues. But I'm not sure that we're having a lot of recommendations or  
45 discussions here about how to change the reimbursement for genetic testing and genetic

1 services. Not only the testing, but the genetic counseling, and other types of services which are  
2 also inadequate.

3 DR. McCABE: So Debra has taken a strong position in support of  
4 reimbursement. Is there any member of the committee that would wish us to take  
5 reimbursement off of the table?

6 MS. BERRY: I think it needs to be on the table, but it is sort of a linear  
7 thing, because it doesn't become an issue if there is no coverage. I mean, they aren't going to  
8 pay for something that's not covered. So the coverage part is the first threshold question, so  
9 reimbursement is by itself, it covers all of the insurers, Medicare, Medicaid, private sector, and  
10 it is critical. It is something that we are tasked with looking at, because it is a significant  
11 barrier to access. So we need to sort of think of them, and I don't know if it is linear, horizontal  
12 or vertical.

13 DR. LEONARD: Well, the problem is that there are CPT codes out there,  
14 and they are generic codes, so you can use those codes for any kind of testing, covered or not  
15 covered. It is not clear to me how insurance companies know what they are paying for, whether  
16 to cover it or not, since the CPT codes are generic.

17 That is going to be remedied somewhat by the CPT code modifier system  
18 that is supposedly coming this fall. But it is not clear until that happens and laboratories start  
19 using those modifier codes as to how much that will decrease denials because of coverage  
20 decisions. They will now know what they are covering or not covering and can say yes or no,  
21 because instead of just saying no because they don't have a clue what the test is, and why it was  
22 done.

23 But that still doesn't address once they decide to cover it, that it is a covered  
24 test, that what you get paid is inadequate for what it costs you to do it. So you're right, it is a  
25 linear process, but I don't think that we cannot go to the next step of at least figuring out what  
26 to do about the inadequate reimbursement, supposing that something is covered, and address  
27 those.

28 You don't really have to address the coverage issues before you look at the  
29 reimbursement issues, because it either is or isn't covered. The reimbursement is inadequate,  
30 even if it is covered. So the reimbursement is a separate issue, and you don't necessarily have  
31 to decide the coverage before you look at the reimbursement concerns.

32 MS. BERRY: Agnes?

33 MS. MASNY: The only place we would need to look at coverage then  
34 would be just the screening issue, because that is where if Medicare doesn't cover screening, or  
35 see screening tests as covered, and I think that was your point earlier about looking to the  
36 commission and agency to define, or commission HHS to allow CMS to incorporate screening,  
37 so that would be a coverage issue.

38 DR HANS: It is actually in my written comments, but I would just remind  
39 the committee that HHS actually does have a health delivery system that it is also responsible  
40 for, and that is through the Indian Health Service. So you actually have within the purview of  
41 this committee, a delivery system.

42 While I don't have any specific recommendations, because I'm not quite sure  
43 where this is going, I would just sort of put a mental placeholder in that as you move toward  
44 more concrete recommendations, that you then think, does there need to be a footnote for IHS?  
45 Just to think through what are the implications for the health delivery system that controls

1 pretty much all the elements that you're talking about here directly.

2 I'm not sure that there would be any special circumstances, but I would just  
3 want to put that on the table for folks to remember. Of course it is rather  
4 self-serving, because if there are things for IHS, there is likely to be also for VA, and we would  
5 therefore find that recommendation is more helpful for us.

6 DR. McCABE: The cultural sensitivity issue is certainly one for IHS and  
7 some of the Indian nations, and genetics does not fit into their belief system, and so they really  
8 have decided as a nation, not to go down this road.

9 MS. BERRY: Debra?

10 DR. LEONARD: One of the areas of coverage that may be able to be more  
11 specifically addressed, which Muin had brought up, is when the use of a genetic counselor is  
12 appropriate. We talk about moving to more generic services being delivered as part of all  
13 health care professional services, but maybe if criteria for coverage of genetic counseling  
14 services could be developed, then it would be clear when those could be covered, and when not  
15 necessarily covered. I don't know if those already exist, maybe someone can comment.

16 That would be a separate issue from reimbursement for those services. But  
17 I think defining when as we move toward the 90 fevers that are dealt with by generalists, and  
18 the ten fevers that are dealt with by specialists, maybe we need to distinguish when you would  
19 pay for the special genetic counseling services.

20 DR. McCABE: I would like to move us on, then to the two topics under  
21 broad issues, and discuss inclusion or exclusion of those, and if they are to be included, then  
22 the rationale for including them under coverage and reimbursement of genetic technologies and  
23 services.

24 Neither of them are dealt with in the policy implications, and that is why I  
25 think it is important that we assist staff with this at this point in time. Health disparities, the  
26 way I see that included, is that if there are populations that are not covered, and/or not  
27 reimbursed, then that will lead to a health disparity. But could we have some additional help  
28 from the committee for staff in terms of the rationale for that one being included in a coverage  
29 and reimbursement manual?

30 MS. BERRY: Just to play devil's advocate, I actually regard both of those  
31 issues as being so important as needing to stand on their own. I don't see them in the context of  
32 coverage and reimbursement, but I see them as broader than that. They could lend themselves  
33 to their own reports, each one. So I'm not clear as to how we would fit them into a coverage  
34 and reimbursement report, or if we should.

35 DR. HANS: Let me ask a question and see if you think that it is an issue. If  
36 you set down some criteria for making coverage decisions based on some evidence, what if that  
37 evidence does not adequately cover certain ethnic populations? What then coverage  
38 recommendations do you make for those populations? And is that a health disparities issue  
39 there?

40 DR. LEONARD: I think what you're getting at is the broader issue of the  
41 science disparities by ethnic group that will influence the evidence base that will be use to  
42 make coverage decisions, that will then influence the health disparities. So you have to go all  
43 the way back to the science.

44 DR. HANS: Absolutely, that's the train. But if you are making a policy  
45 decision, and for Caucasians, the test has been validated, what do you then do as a policy

1 entity? Do you say you cover it for anybody, but the evidence doesn't exist for the other ethnic  
2 populations?

3 I don't know how you begin to help policymakers figure out how to make  
4 those decisions. Or do you wait until you have evidence that covers all ethnic populations  
5 before you make a coverage decision?

6 DR. McCABE: How about provider education and training, then? I think  
7 the argument has been made that we do need the evidence base for populations if we are to  
8 have recommendations. What about provider education and training?

9 Again, and I understand Cindy's point that these are huge topics. But if we  
10 focus them, I think we can focus the topic and take one little piece of it for coverage and  
11 reimbursement, not try and deal with all of health disparities, but just as they apply to this  
12 topic. Would that be then more reasonable, Cindy?

13 MS. BERRY: Then how does provider education relate to coverage, for  
14 example? We know all the issues about the inadequacy of the education for our providers and  
15 the gaps and public awareness and all of that, but how does that relate to coverage? Emily?

16 DR. WINN-DEEN: I want to make a comment on health disparities, and  
17 then I'll answer your question. I think the other thing that we have to do, besides setting up  
18 evidence-based studies that take into account at least the three major branches of the human  
19 tree, and make some kind of statement about whether a test is or isn't appropriate for each of  
20 those branches, let alone all the twigs and leaves that are off of those branches.

21 We also have to recognize the economic disparities that exist, and whether  
22 there is a group of people who are willing to self-pay, and thus gain access to things earlier,  
23 while coverage decisions and evidence-based medicine are churning along, and whether that  
24 creates some additional health disparities or access issues, however you want to put it.

25 I think there is a sort of genetic group disparity where we have to deal with  
26 that, and we set up evidence studies. But there is also the economic disparities that people who  
27 have the means will pay for almost anything. To what extent do we feel as a policy group we  
28 need to address that? And at what point are people spending money on things for which there  
29 isn't evidence? Is that just their own buyer beware, people pay for a lot of snake oil, and only a  
30 little bit of it works?

31 So that's one thing. The other, in terms of provider education and training, I  
32 think that again, comes down to potentially an issue of disparities, where if you are seeing  
33 somebody at a highly acclaimed university medical center, you might get a different level of  
34 provider education than if you are seeing someone in a rural outpatient clinic. It just depends  
35 on where you live, and the kind of training that the people who you are seeing may have  
36 received.

37 So again, you have the issue of getting equal access, equal treatment  
38 availability to everyone, without having equal provider ability to deliver that.

39 MS. BERRY: My point is that I agree with everything you're saying, and I  
40 think of these issues as being at the core of the overall access umbrella that we've been  
41 struggling with, and I think they do merit further attention, and maybe their own report. But I'm  
42 still not clear on how they directly relate to coverage and reimbursement.

43 Not that I think that they aren't important, but I want to focus the report on  
44 those two issues, since that is what it is supposed to do, and then address these others, which  
45 we have to get our arms around in a broader context, because it cuts beyond those two issues of

1 coverage and reimbursement, I think.

2 Debra, and then Martin.

3 DR. LEONARD: Well, the health disparities, I think directly is related to  
4 coverage and reimbursements since they are different systems, and they are uninsured and  
5 underinsured. If Medicaid is decided state by state, it may be harder to influence that coverage,  
6 and so that may be less adequate coverage than something that can be influenced nationally.

7 So the way coverage and reimbursement is done does create health  
8 disparities, and maybe we should look at those issues. The provider education and training,  
9 aren't we doing that in the other resolution? I don't really see how that affects coverage and  
10 reimbursement, since coverage and reimbursement is decided on an evidence base that is not  
11 related to whether physicians know how to use tests appropriately.

12 So in my mind, that broader issue is really more addressed in the other  
13 resolution that we discussed this morning, and not so much a piece of the coverage and  
14 reimbursement.

15 MS. BERRY: Martin?

16 MR. DANNENFELSER: Yes. It would seem right now that if people have  
17 the economic means to pay for things out-of-pocket, they are going to pay for them. So the idea  
18 that we wouldn't provide coverage because somebody else might get it, I don't see the issue  
19 there. I think that people of lesser means are not going to have the capability now. So if you  
20 make it available, make the coverage available, then you are providing them the means, and you  
21 are increasing their access.

22 So I think we can get to the point of trying to micromanage or letting, as  
23 was said before, let the perfect become the enemy of the good, if we're going to say that people  
24 don't have access now, then we're going to provide them access, we're enhancing, we want to do  
25 what we can to make sure that the level of training and services are equitable, or as close to  
26 equal as possible. But I think if we wait until everything is perfect, we'll never be able to act.

27 MS. BERRY: Barbara, did you have a comment?

28 MS. HARRISON: I basically wanted to echo exactly what Debra said. I  
29 can definitely see the link between health disparities and how that needs to be addressed in our  
30 report, because what comes out of our recommendations about coverage is going to directly  
31 affect the access that people have to genetic services, and I think this point has been brought up  
32 before, not just talking about disparities among ethnic groups, but also economically. If we're  
33 talking about things like Medicare, Medicaid, and what it is covering, then to me, it is just very  
34 plain about how that affects health disparities. So we can have that be a continual theme  
35 throughout the report as to why this is so important.

36 But again, agreeing with previous comments on the provider education  
37 piece, I don't necessarily see it being directly affected by our report on coverage and  
38 reimbursement. Definitely to access to services, but not in this report.

39 MS. BERRY: Emily?

40 DR. WINN-DEEN: Yes, so I guess I think the only place it belongs in the  
41 report is at the end where we say just having coverage and reimbursement does not assure that  
42 this becomes practice of medicine, that there are still other barriers that we have to deal with.

43 So that is where I'd just like to see sort of a sentence at the end that says,  
44 this is a huge barrier. If we overcome coverage and reimbursement, we are 90 percent of the  
45 way there in delivering this promise of genetics to the practice of medicine. But there still are

1 some other things that have to be in place for that to really be totally efficient. That's the only  
2 role that I see in it in this report.

3 DR. LEONARD: And can't we reference the other resolution?

4 DR. McCABE: Yes. I think it is always good to cross-reference ourselves,  
5 and I think it is also important to, and I think perhaps in that paragraph we could use some of  
6 the data that Reed provided us with that shows that even where there are U.S. Preventive  
7 Services Task Force guidelines, they are still not fully utilized. He has some excellent data in  
8 his comments that he sent to us.

9 Well, I want to thank everyone for a very productive discussion, and for a  
10 very productive day. I want to especially thank Dr. Boughman, Dr. Reede, Ms. Berry, and all  
11 of our panelists and public commentators.

12 We are now going to adjourn for the day. Members who are planning to  
13 join us for dinner this evening, please meet in the lobby at 6:40 p.m. so that we can then  
14 carpool to dinner.

15 To remind everyone, we are starting a half hour earlier tomorrow, so we  
16 will reconvene at 8:00 tomorrow morning. Thank you, and we're adjourned for the evening.

17 (Whereupon, at 5:22 p.m., the meeting was recessed, to reconvene at 8:00  
18 a.m. on Tuesday, June 15, 2004.)  
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