U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

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

Fifth Meeting

Tuesday,
October 19, 2004

Congressional Ballroom
Bethesda Marriott
5151 Pooks Hill Road
Bethesda, Maryland

IN ATTENDANCE:

Chair

Reed V. Tuckson, M.D.
Senior Vice President
Consumer Health and Medical Care Advancement
UnitedHealth Group
MN 008-T902
9900 Bren Road East
Minnetonka, MN 55343

Members

Cynthia E. Berry, J.D.
Partner
Powell Goldstein Frazer & Murphy
1001 Pennsylvania Avenue, N.W., 6th Floor
Washington, D.C. 20004-2582

Barbara Willis Harrison, M.S.
Certified Genetic Counselor and Instructor
Division of Medical Genetics
Department of Pediatrics
Howard University College of Medicine
Box 75, 520 W Street, N.W.
Washington, D.C. 20059

Debra G.B. Leonard, M.D., Ph.D.
Vice Chair of Laboratory Medicine
New York Presbyterian Hospital, Cornell Campus
Room F715, Mailbox 79
525 East 68th Street
New York, NY 10021

Brad Margus
Co-Founder and Volunteer President
A-T Children's Project
Co-Founder and CEO
Perlegen Sciences, Inc.
2021 Stierlin Court
Mountain View, CA 94043

IN ATTENDANCE:

Agnes Masny, R.N., M.P.H., M.S.N.
Adjunct Assistant Professor of Nursing
Temple University College of Allied Health Professionals
Research Assistant and Nurse Practitioner
Family Risk Assessment Program
Fox Chase Cancer Center
7701 Burholme Avenue
Philadelphia, PA 19111

Edward R.B. McCabe, M.D., Ph.D.
Professor and Executive Chair
Department of Pediatrics
David Geffen School of Medicine at UCLA
Physician-in-Chief
Mattel Children's Hospital at UCLA
10833 Le Conte Avenue, 22-412 MDCC
Los Angeles, CA 90095

Huntington F. Willard, Ph.D.
Director
Institute of Genome Sciences and Policy
Vice Chancellor for Genome Sciences
Genome Sciences Research Building II
101 Science Drive, Room 4011
Duke University Medical Center
Durham, NC 27708

Emily S. Winn-Deen, Ph.D.
Vice President
Strategic Planning and Business Development
Cepheid
904 Caribbean Drive
Sunnyvale, CA 94089

Kimberly S. Zellmer, J.D.
2525 Tomahawk Road
Mission Hills, KS 66208

Ex Officio Members

Administration for Children and Families

Martin Dannenfelser
Deputy Assistant Secretary for Policy and External Affairs
Administration for Children and Families
370 L'Enfant Promenade, S.W., Suite 600-W
Washington, D.C. 20447

IN ATTENDANCE:

Agency for Healthcare Research and Quality

Francis D. Chesley, Jr., M.D.
Director
Office of Extramural Research, Education,
and Priority Populations
Agency for Healthcare Research and Quality
540 Gaither Road
Rockville, MD 20850

Centers for Disease Control and Prevention

Muin J. Khoury, M.D., Ph.D.
Director
Office of Genomics and Disease Prevention
Centers for Disease Control and Prevention
4770 Buford Highway, MS K-89
Atlanta, GA 30341

Centers for Medicare and Medicaid Services

James Rollins, M.D.
Centers for Medicare and Medicaid Services
7500 Security Boulevard
Baltimore, MD 21244-1850

Department of Defense

Colonel Martha Turner, USAF NC, Ph.D.
USAF Surgeon General's Consultant for Medical Ethics
U.S. Department of Defense
Preventive Medicine and Biometrics
International Health Specialist Program
Uniformed Services University
4301 Jones Bridge Road
Bethesda, MD 20814

Department of Justice

Vahid Majidi, Ph.D.
Chief Science Advisor
Office of the Deputy Attorney General
U.S. Department of Justice
950 Pennsylvania Avenue, N.W., Room 4217
Washington, D.C. 20530

IN ATTENDANCE:

Department of Veteran Affairs

Sherrie Hans, M.D.
U.S. Department of Veteran Affairs
810 Vermont Avenue, N.W.
Washington, D.C. 20420

Equal Employment Opportunity Commission

Peter S. Gray
Senior Attorney Advisor
Office of Legal Counsel
U.S. Equal Employment Opportunity Commission
1801 L Street, N.W.
Washington, D.C. 20507

Food and Drug Administration

Steven Gutman, M.D., M.P.H.
Director
Office for In Vitro Diagnostic Device Evaluation and Safety
Food and Drug Administration
2098 Gaither Road, MSC HFZ 440
Rockville, MD 20850

Health Resources and Services Administration

Suzanne Feetham, Ph.D., R.N., FAAN
Senior Advisor
Office of the Director
Bureau of Primary Care
Health Resources and Services Administration
4350 East-West Highway, 11th Floor
Bethesda, MD 20814

National Institutes of Health

Alan Guttmacher, M.D.
Deputy Director
National Human Genome Research Institute
National Institutes of Health
Building 31, Room 4B09
31 Center Drive, MSC 2152
Bethesda, MD 20982

IN ATTENDANCE:

Office for Human Research Protections

Michael A. Carome, M.D.
Associate Director for Regulatory Affairs
Office for Human Research Protections
1101 Wootton Parkway, Suite 200
Rockville, MD 20852

Executive Secretary

Sarah Carr
Secretary's Advisory Committee on
Genetics, Health, and Society
6705 Rockledge Drive, Suite 750, MSC 7985
Bethesda, MD 20892-7985

C O N T E N T S

C O N T E N T S

P R O C E E D I N G S (8:35 a.m.)

DR. TUCKSON: I want to welcome everyone to the second day of our meeting. By the way, right at the outset, public comments are always requested and happily received. If there is anyone that wants to sign up for the public comment time, you just go over to our ace people here at this table. Actually, if you go this table here, they'll direct you to the table outside, and you can sign up. It depends upon how simple you want to make it.

We got a lot done yesterday. We got, I think, very serious about six or seven initiatives which we will review for you when we get through to near the end of the day on the anti-discrimination issues. I think we've got to stick to that level of focus and serious recommendations today if we are to have an overall successful meeting. So we're going to have to really think very carefully about what to do with the remaining recommendations in this report on coverage and reimbursement, and then on population.

One thing that I will ask Sarah and her team to give out, which we'll do probably at the break and put in front of you at the break, would be a little flow chart of all of the things that we have on our plate right now, and some things which look like they're supposed to come onto our plate, and sort of what they look like from a time sequence point of view. It is like a big picture view of all the stuff.

The reason why I want that for us to have around the break or so is as we get near the end of the day, I think we have to start to look very carefully at again, focusing on how much we can bite off, exactly again what we are intending to bite off, and how we can turn each of those into definite win projects for us.

So I just want to make sure we don't get overwhelmed and not really see what we have in front of us. So we'll give you that around break time.

We welcome the people that are watching by webcast, if they are in fact watching. Is that on?

DR. McCABE: Yes. Yesterday, they were saying we had 35 to 40.

DR. TUCKSON: And I do know from emails, as I said, they were mainly talking about Ed's tie, but they were there. So they are there. It was inconsequential, I heard. But we thank everyone who is on by webcast, and we really do appreciate this way of reaching out to the public.

With that, let's go back to Cindy Berry and take up where we resumed yesterday.

MS. BERRY: Everyone should have the handout that was distributed this morning. The front page goes over what we at least temporarily tentatively decided in terms of recommendations with regard to the screening exclusion and the national versus local coverage decisions. I won't go over those again, but take a look. The reverse side addresses genetic counseling. We started that, but didn't wrap it up.

We went back with staff and came up with a revised set of potential recommendations, and wanted to start this morning going over that. The first recommendation talks about the analysis that we discussed. We didn't specify who would do the analysis. The analysis could be simply reliance on what HRSA is already doing. If not, it could be the Institute of Medicine. We are in the process of trying to determine the state of play of the HRSA effort, and determining whether an additional analysis is needed. So we have purposely left that vague.

The idea is that someone needs to do a comprehensive look at who is

qualified to provide genetic counseling services, under what conditions, who should be supervised, who doesn't need supervision, and how should they be reimbursed. It should also look into the effectiveness and value of providing these services.

If there are any gaps in that analysis that are uncovered, that could lead to a Medicare demonstration project which would provide us with additional data that would help address some of the questions and barriers outlined in the report on genetic counseling access. The third proposed recommendation has to do with a legislative change.

If the data analysis and the data gleaned from the demonstration project indicate and support this, and we all think it will, then perhaps we would be in a position to advocate a legislative change, a congressional fix where we would add all of the appropriate health care providers to the list of non-physician providers who can directly bill Medicare.

We purposely left that open, not really wanting to limit it to one specialty or another, because there are many different health care providers who are able and capable of providing these types of services. So the analysis and demonstration project, if we have one, will help inform who those people should be.

The fourth potential recommendation addresses the issue of licensure. Licensure of genetic counselors in particular. We have a question mark there, because the feeling was that we'd probably need a little bit more discussion on that. Is this something that this committee should promote? Do we have any authority, or does the Secretary have any ability to influence states in whether they set up some sort of licensure process for different health care providers such as genetic counselors? That's a question mark in my mind, and I'd welcome further discussion. I wish Barbara were here, because I know she'd have some input on that as well.

Ed, and then Debra.

DR. McCABE: I think it's certainly a critical piece, because without licensure, it is going to be difficult to bill. I think you are also correct that it is going to be hard for us to have an impact, because this is a state by state issue.

It can certainly be built into the logic of the argument that it is a critical piece. I think we should try and recommend things that are doable, even if they are a bit of a reach. I think this one is probably not really doable, but should certainly be commented upon in the logic of the argument.

MS. BERRY: Do you think, Ed, just to follow up, that perhaps the issue would be addressed in the meat of the report, but not constitute a recommendation? Or do you have a thought for how we could phrase a recommendation that would address this?

DR. McCABE: I think it should be definitely in the meat of the report. It could be under Number 1, because that addresses, if we look at the first bit, an analysis that will determine which health providers are qualified to provide genetic counseling, under what conditions, and under what supervision. I think it is the supervision point that if there is licensure, then there is less need for supervision, so that one could build it into that first sentence, perhaps, in a parenthetical.

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

DR. LEONARD: I know that genetic counselors are very, very valuable today in the services that they provide. They can't bill, and they can't be paid for what they do. These services are underwritten by departments just because it is a necessary part of medical genetics.

I feel like only asking for a study, although maybe necessary is leaving those genetic counselors who are out there practicing still flapping in the wind. I don't know if that is a very effective thing to be doing. If I was a genetic counselor looking at these recommendations, I'd probably be very angry, and feel like I wasn't getting any assistance from this committee, because a study is going to take at least six months to be commissioned, and about a year to 18 months to do. There are people out there currently practicing and adding a lot of value in the medical genetics area.

MS. BERRY: Do you have a specific idea on how you might address that, or some of their concerns? Is licensure where you would put the focus? Or are there other areas that you think we should focus on?

DR. LEONARD: Well, there is licensure coming in two states, so there is a move towards licensure. Maybe we should create a mechanism whereby people that are licensed to provide genetic counseling services can be acknowledged as allied professionals and get a UPIN so that they can bill for the services that they are providing as licensed health care professionals.

MS. BERRY: Emily, Muin, Hunt, and then Ed.

DR. WINN-DEEN: So I think we also heard pretty strongly from Barbara yesterday, so I'll just try and represent this again, that there is already a national accreditation program for genetic counselors. I, for one, have a little trouble understanding why a nationally accredited counselor also needs to go through a state licensure, and why that national level of accreditation isn't acceptable per se.

Now, I understand you might have to send the state you live in $50 and get a piece of paper. But I think we also ought to encourage that people who have received that level of national accreditation on the basis of a Master's degree in genetic counseling, that that ought to be sufficient as well.

DR. LEONARD: But it's not in any health care profession. So physicians who have gone to medical school, residency, training, board certification, and everything else, can't practice unless they get a license from the state in which they practice. So this is standard for being able to bill. So I do think you can't get around the licensure. That is going to have to be done on a state by state basis.

If there are states that are licensing, then if we allow those states to bill, maybe it would encourage other states, not those states to bill, but genetic counselors in those states to bill, because they are licensed and can get a UPIN and bill, then maybe there would be other states that would license as well.

MS. BERRY: Muin?

DR. KHOURY: I would like to urge the committee not to be a bit myopic in its view. Right now, I think you are just focusing with a real tunnel vision on the specialty of genetic counseling. I mean, if you think about the practice of medicine in the next 10, 20, 30 years, there is going to be the delivery of genetic information not only for the diagnosis and management of genetic diseases which account for 5 percent of all human disease, but in the management, prevention, and diagnosis for all diseases, whether they are genetic or not.

So if you kind of step back here and you want to provide the maximum value added advice to HHS, then ask the question, how are we going to be ready in delivering genetic information? I'm not saying genetic counseling is not important, because genetic counseling is of paramount importance, at least in a fraction of the delivery of genetic

information.

What has happened over the years is that genetic counseling as an institution has really grown up to have a very well defined way of doing business primarily in a non-directive fashion, or the usual way of delivery of genetic services. Whereas the new practice of medicine in the 21st Century is going to be dictating a new paradigm shift in how information is going to be delivered.

For a fraction of cases or people who come through the clinic, the traditional genetic services paradigm will still hold. Increasingly, we are going to be faced with applications from pharmacogenomics, somatic cell genomics, and gene expression arrays for which information is not going to be necessarily delivered through the genetic counseling route.

So, for example, instead of saying an analysis is needed that will determine which health providers are qualified to provide genetic counseling, if you kind of expand your look and you say, of course we're expecting health care providers to provide genetic information in the future, but how do we get there? Under what consensus do we rely on genetic counselors to do that part of the work?

To me, the practice of genetics in the 21st Century is going to be a lot like the practice of infectious diseases, because right now, anybody can order a blood culture. Any health care provider can order a culture and can prescribe antibiotics. So what is the infectious disease specialist? The role of the infectious disease specialist is probably to teach the health care providers, what did I think to do? Maybe in a small subset of complicated or complex cases, to take them on, to be consulted on.

So I would urge the committee to have a more open view of the practice of medicine in asking the genetic counseling questions under the umbrella of delivery of genetic information. Because once you open that door, genetic counseling will have a role in it, but it won't be the only thing that you will focus on.

MS. BERRY: Hunt?

DR. WILLARD: I can't add much to Muin's typically articulate phrasing of the issue. I had scribbled down many of the same issues in terms of trying to broaden this to genetic and genomic services of which genetic counseling is clearly one avenue, rather than carrying out what I heard Debra say, which is a sense that our remit here is to try to help and rescue one particular subset of the workforce. Although any of us as individuals may bring that to the table, I don't think that that is a particular charge to the committee.

As Muin said, it is a charge to the committee to advise the Secretary on how best to organize the provision of genetic and genomic informational services. To me, licensure issues are way, way down the list. Especially if one tries to create a linkage to billing. Licensure of physicians, they are not getting a license to bill. They are getting a license to practice medicine even if they weren't billing, even if they were doing it for free, they'd still have to have a license.

So there is no direct connection between the act of licensure, and the right to bill. I would sort of pull ourselves up as a committee to the sort of 5,000-foot level of how do we best organize the scene to provide services globally for all the groups that we can imagine might be involved in providing genetic and genomic services, of which board-certified genetic counselors are one, but by no means the only group that will be relevant to that.

MS. BERRY: Ed?

DR. McCABE: To move us forward, I have written down some things here,

taking the comments and some of my own thoughts. We could relabel this section on genetic services and counseling. We could take the first sentence and say, "SACGHS believes that delivering genetic services, including genetic counseling, is a critically important component."

We could add a sentence at the end of Number 1. I understand Debra's concern about analysis, but I think unfortunately it needs to be deliberate. It may be that these analyses are underway, and therefore, that would speed things up. I would add a sentence at the end of Number 1, "This analysis should address workforce needs, independent practice and licensure of health providers delivering genetic counseling, and other genetic services."

So to try and broaden it out. I know it is not as broad and not as high an altitude level as was recommended, but I think it does broaden it a bit and gives us some specific language.

MS. BERRY: Emily?

DR. WINN-DEEN: Well, okay, so I think that we should make sure that if we're going to broaden it beyond genetic counseling, right now this box sits in a section entitled "Genetic Counseling," which I think, Hunt, is why it was limited to genetic counseling.

One thing that I think we haven't captured in this box that we discussed to some extent yesterday was the provision of counseling by other individuals then officially designated genetic counselors, such as oncology nurses, and such as practice nurses who have specialized knowledge of a disease area where they are working. A cystic fibrosis clinic or whatever kind of a disease specialty area.

I think we shouldn't ignore, and I think this is maybe where Hunt was coming from. We shouldn't ignore all these other people that are doing this today, providing counseling services without the official imprimatur of being a genetic counselor. Also the fact that as we move into the genetics of common complex diseases, that this will have to be a much more distributed effort. There is no way we could concentrate it in a single small subspecialty.

MS. BERRY: Ed?

DR. McCABE: That's why I broadened it to say genetic services including genetic counseling. I think that would allow one to address the other health providers and evaluate what the roles of these individuals could be.

DR. LEONARD: And I think it was mentioned yesterday also that the nurses who are providing these genetic counseling services are certified. So there is a certification process that says they are qualified to do these things.

DR. WINN-DEEN: I'm not in any way saying that the people who do it shouldn't be qualified. I'm just saying that there are people who are not designated officially as genetic counselors who are doing it. They have received the training, the knowledge, and they are prepared to do it. Some of them are physicians, some of them are nurses, and some of them are probably social workers in some cases.

MS. BERRY: What we tried to do yesterday was to broaden this, because there was some sensitivity about limiting it to one specialty, or one area of practice. But Ed's changes, I think, and with Muin's recommendations, really improve upon that.

Perhaps then to address Emily's point, we beef up the substance of the report, because I don't think there is enough discussion of all of the different specialties. It was initially drafted as a genetic counseling piece.

So as you pointed out, Emily, we need to make the substance fit in with the recommendation, so to speak.

DR. TUCKSON: Could you reread then the last draft that we got from Ed?
MS. BERRY: From Ed?
DR. TUCKSON: And/or with the Muin modification.
MS. BERRY: The section then would be entitled "Genetic Services and Counseling" or it could be "Delivery of Genetic Services and Counseling." "SACGHS believes that delivering genetic services, including genetic counseling, is a critically important component of the appropriate use and integration of genetic tests and services." We might add in there, "into the practice of medicine, our delivery of health care."
Then the first recommendation is as it appears up there, but adding a sentence. I do actually think we need, because it references only genetic counseling, so we'll need to change that to talk about genetic and genomic services, or some other broader term. Then add a sentence that would read, "This analysis should address workforce needs, independent practice and licensure of health providers delivering genetic counseling, and other genetic services."
So the idea of being where genetic counseling is mentioned specifically, that could be included in a component, but the broader terminology which would be something like "delivery of genetic and genomic services" would be inserted in its place.
DR. TUCKSON: All right. So if I understand it then is that what happens here is that we are saying that this whole field, there are a number of interventions, whether they are counseling and/or procedures and/or other things, that are related to this new field that require clarity around qualifications, scope of practice, so forth and so on. And that that work has to occur, and that we are calling for some order leading us to that.
Then the link is that in absence of that, it is very difficult to recommend reimbursement for those services, given that you don't have this fundamental predicate well enough established or in place. So we are calling attention to the need to be able to create clarity or guidance around reimbursement, but we are making it clear there is a predicate step that has to occur first. Is that essentially what we're doing here?
MS. BERRY: I think you've captured it.
DR. TUCKSON: And then, I guess the second question would be is there any particular reason that we should from that make a special mention of genetic counseling and the reality today for that particular domain. So okay, we have said this now for the whole skrabish.
By the way, right now, given that there are not a lot of interventions to talk about, but right now there is a lot of counseling going on, do we need to, for the sake of the counseling community, sort of say there needs to be some prioritization around moving this along for the counseling community?
DR. McCABE: I think that we actually say genetic counseling, and we put it in small letters rather than in caps. But it is definitely there in several places.
I think what it does is it recognizes those who are doing genetic counseling, which clearly would then be in capital letters, the genetic counselors, the masters, board-certified genetic counselors. But I think also the intent of my amendment was to also address the other individuals who are providing genetic services, including genetic counseling.
DR. TUCKSON: The thesis of my second point is simply to recognize I guess a sort of, and again, I'm just throwing it out there, is the recognition of a certain urgency and prioritization that says look, we've got to get all this stuff done, but right now we really got

this real immediate mess on our hands that we need to have resolved. So we are sort of saying as a first priority, there would be these people that we want to get looked at. I don't know whether philosophically that is where the committee is or not.

MS. BERRY: Suzanne?

DR. FEETHAM: To be consistent with the language we have been talking about about services, framing it as genetic counseling services I think keeps that at that level that we've been now revising this whole thing to look at services. Just in the language of again, not going to individuals, but keeping it at a genetic counseling service. I think that encompasses all of the issues we've been talking about.

MS. BERRY: Debra?

DR. LEONARD: I agree with you, Reed, that we do need to address the genetic counseling that is going on now as an urgent issue. I was in no way saying that genetic counselors were going to be providing everything in the future.

But we do know that there is an education gap in the medical workforce. Those who are trained in genetic counseling, be they official genetic counselors, nurses, or whoever, who have gotten the training in genetics will be facilitating the integration of genetics into health care, helping those who don't know genetics to learn that.

Just a clarification. Licensure does not allow you to bill, but without a license, you can't bill. So you do have to have that step before you even have the possibility of billing.

MS. BERRY: Does anyone have any further comment on the three recommendations as revised?

DR. LEONARD: Are we going to add a fourth on the lines of what Reed suggested?

MS. BERRY: Suzanne, have you captured ? -

DR. KHOURY: I have another idea. One of the things that got deleted from yesterday is calling for an IOM analysis of the effectiveness of genetic counseling. This is one of the kind of sticky points right now in the practice of genetics because of the lack of a billable entity related to genetic counseling where people spend a lot of time imparting information that could be useful to people and their families.

I think if we are to call for sort of the big picture recommendation as Number 1 related to the delivery of genetic information and services in general, I think we owe it to the practicing community right now to kind of evaluate in sort of broad terms the ? - I mean, the way you couch it here is effectiveness. Of course people who are practicing this specialty will say, of course it is effective. We are imparting information that would be useful to people.

But when I have had many discussions with a lot of my friends in genetic counseling, what seems to be lacking is a lot of outcome research that could be measurable in a sense to show the clinical utility of that information. I think as a group, I mean, genetic counseling as an entity could benefit from a closer look as to the value and utility of that.

So in other words, if you are to do a randomized clinical trial today, which I don't think anyone will do where you have people coming in and you impart information on with or without genetic counseling, I mean, you can give them a diagnosis and you can send them home, or you can spend an hour or two on genetic counseling. No one is going to do that study for ethical reasons.

But if that study cannot be done, therefore genetic counseling and that hour or two hours should be billable. It has imparted useful information. So it is kind of is a Catch 22 with respect to delivery of current genetic services for single-gene disorders.

I think a closer look as to the utility of that approach is useful. Maybe somebody has done that sometime, but I'm not aware of it.

DR. McCABE: I just wanted to comment.

Muin, you said that it wouldn't be ethically appropriate to do it. Actually, there are some trials ongoing right now with pediatric surgery looking at surgical procedures and clinical trials, which is almost unheard of in surgery to actually look at the efficacy of various surgical procedures. So I think one could design a trial that would be acceptable to IRBs and would answer these questions.

MS. HARRISON: This is Barbara. I think there are also studies that are out there that show different ways to do counseling, whether the counseling is done by a physician versus a trained genetic counselor, and some outcomes from that kind of data. So there are some studies out there that show the efficacy of counseling. I think a literature review effort could unearth some of those.

Otherwise, I just want to make sure, and I think it is getting in there now that I guess my two key issues are licensure to make sure that we put our support behind that being done, as well as I think identifying genetic counseling separately as a key point.

DR. TUCKSON: I was asked to put my thought up there. So what I did was it is actually in Number 1. I don't think it is a separate 4. I think at the very end of Number 1, if we were to say in this next to the last sentence, "This analysis should also address workforce needs, independent practice, and licensure of health providers delivering genetic services. The committee urges that genetic counseling services be a priority for this activity."

I don't know whether that helps or not, but at least you get the whole gamish. Then we come back and say that genetic counseling is a priority.

DR. FEETHAM: As I mentioned yesterday, just a reminder of the study funded by HRSA that was led by Dr. Judith Cooksey is ending at three years. Again, the findings of that may inform. So I just want to remind. I don't have those findings at this point in time, but I just want to remind you that that has been going on, and that may help inform this discussion. You are not starting from a blank slate if you are recommending another study.

DR. TUCKSON: And I didn't get to hear all of Muin's comments. But I think in the sense of the urgency and so forth, I think we need to be clear as a committee in how serious are we about pushing this recommendation forward?

We say we're going to do this study or call for the study, or push forward on this. This is a major, I think, take home issue, and we need to decide if at the end of the day, this is going to be one of the top priorities that comes out of this meeting or not, or whether it is just enough that we put it in our report and send it forward to the Secretary. Or do we really want to come back and consider this to be one of the take homes that define whether our committee was a success or not.

I'm listening for my colleagues to sort of give us a sense of once it is in the report, is this an evaluative issue for our committee.

MS. BERRY: Suzanne, I wondered if I could ask, is the HRSA study limited to allied health professionals? Or is it the whole waterfront? We were talking just a moment ago, does it apply to physicians and geneticists?

DR. FEETHAM: It was looking at genetic specialists, including physicians and non-specialists.

MS. BERRY: Okay. Does the committee have any views on whether we should be focusing on that broadly, is that too broad a focus, or should we hone in a little bit more on the allied health professional world?

Right now it is very broad, and it encompasses, the way it is worded, all specialties, all professions that deliver or potentially could deliver genetic services, including physicians.

MS. CARR: I mean, I was wondering about this as well. I mean, the committee is not interested in asking for an analysis of providers that are now licensed, are we? It is only those who are not currently licensed, or whose license is within a certain scope and may need to be broadened. This wouldn't include M.D. geneticists, would it? It would.

PARTICIPANT: (Inaudible.)

MS. CARR: I know, but I'm wondering if we intended to mean that. What are the issues there that we're trying to get at, I guess?

DR. WILLARD: I mean, I think the issue, and Barbara raised it as well, which health providers are best suited to provide the best genetic or genomic health care? In some cases the answer will obviously be physicians, in other cases it may be genetic counselors, and in some cases it may be nurse practitioners. That's how I read that was sort of a very broad look at the entire landscape of genetic and genomic services, and then saying okay, who is lined up to do the best job under the best circumstances?

How is licensure relevant to any of those determinations? That's how I read that.

MS. BERRY: Ed, and then Emily.

DR. McCABE: And that's why I thought it is important, again, to be deliberate and analytical, and develop an evidence base. I think there is some literature there, but I think as we move forward, there needs to be a larger literature to really address the workforce needs.

I think buried within that are issues that we've addressed before in this committee. That has to do with education. Are we educating our physicians? In fact, in the management of genetic disease, and I agree with Muin, I think it is going to diffuse throughout all of medicine. But are our medical students being prepared for that medicine?

That's why I think we need to be analytical. I don't think we need to be deliberate for the next two decades. I think we really need to develop the appropriate evidence base to justify the recommendations that many of us would make from our gut in terms of who does what job better.

When Barbara says there is evidence comparing physicians and genetic counselors, that is a no-brainer as a physician, because it is clear that genetic counselors do a far better job in my experience. I think that literature needs to be reviewed, analyzed, and made more public.

DR. WINN-DEEN: So we're working on something that is focused sort of as an overarching subject for this discussion on coverage and reimbursement. So a lot of these issues that we're talking about are more broad, workforce preparedness issues, if you will.

But I think what might be helpful is if staff could put in this a little box chart that just has who are the allied health professionals that are delivering genetic counseling today, and what is their current status? Are they licensed? Who is reimbursable? Who is not?

That might make it very clear where we need to focus any further work.

Obviously physicians are already licensed, and they can already bill for their time. So that might not be where we need to focus services, although some of those physicians might need more education so they can do a better job of doing the counseling. But from a coverage and reimbursement point of view, I think we need to just sort of remember where this section is appearing, in which report, and not get too far off the focus of that issue.

I think we're trying to address the barriers to service provision, and we want to make sure that the people who are actually doing the services can get paid for their work, so that they are inspired to come in and continue to do that every day.

MS. BERRY: Muin, I thought I remembered you expressing some concern about the line where it talks about assessing the effectiveness. Is there a different wording that you propose?

DR. KHOURY: See, what Emily was trying to tell us is to focus a bit on the scope of this section in the report, which is about coverage and reimbursement. If we live in a world where genetic services and genetic counseling is not covered or reimbursed, the question to ask is why.

I mean, if these people, whoever they are, are providing services that are useful to individuals and families, why can't we analyze to see what kind of services we would lose if these services are not provided. If we lose those services, and the health outcomes or psychological outcomes are so much worse off, then why aren't we paying for them?

So it seems to me, and I don't live in this world. I mean, if people say that there is outcomes research out there, let's put it together. This committee can actually recommend that this can be done. We can do it between now and the next meeting and make those recommendations stand better on their feet.

I agree. This is not about the education of the workforce, because that is not in this report. It is about coverage and reimbursement. So given the diffusion of genetics and all of medicine, right now today genetics is concentrated on genetic diseases. So we have to at least solve that part before we begin to diffuse genetics in all of medicine.

I think that the first part, which is single-gene diseases, hasn't really been sold in terms of coverage and reimbursement. So I think this committee can make a very incisive recommendation to at least address and assess these issues.

MS. BERRY: Reed?

DR. TUCKSON: Just a subtle addition, and it doesn't change any of the flavor of what Muin said. But I just noticed that we have not said, at least I have not heard that we said explicitly that one of the tasks of this is also to make sure that people don't get ripped off. There is a protection of the public in all of this as well. We focused all of this on what are the good things.

Somebody sets up shop on the corner and says, you know, you've got a genetic disease, I'll talk to you about it. Insurance won't pay for it, but I only charge $30 an hour. It is like H&R Block or something like that. Or telephone hotlines that you know are already there and are springing up.

So we have a telephone hotline thing, and we take credit cards. How does the public know whether these people are any good? So it is part of the reimbursement issue as well. I just want to add that perspective to it.

MS. ZELLMER: Muin, I would also say that for single-gene disorders,

there are a lot of people who don't get genetic counseling who would benefit from genetic counseling. Most of their information comes from their physician, and even, you know, some of the specialized physicians are not very good at giving genetic advice.

I think that perhaps the coverage and reimbursement issue restricts people from getting genetic counseling, and I think a lot of people get their information over the Internet or from whatever they can do to find their resources.

I think that certainly we should do whatever we can do to encourage people, particularly with the single-gene disorders, and perhaps as we move on talking about genomic medicine, hopefully we can worry about the education component and get probably more physicians involved in the counseling process.

I certainly think with single-gene disorders, I think there is a large percentage of the population who does not get genetic counseling. Probably I would guess because genetic counseling is a paid-for service, and it is not getting recommended by physicians.

MS. BERRY: I think we've got some work to do in terms of framing this issue more broadly and incorporating all the comments that we heard this morning. We'll do that. There is some work that was done by SACGT that I think we can lift from and teeing up the issue that will help put context to these recommendations.

So if I can kind of bring us to a close on this issue, understanding the points that everyone had raised, and accepting the fact that we are going to work on language in the substance of the report that provides that level of explanation and context.

Does everyone feel comfortable with these three recommendations as they are currently presented? Any objection? Reed?

DR. TUCKSON: I was just trying to make sure. So an analysis is needed ? - I'm trying to see.

MS. BERRY: It could be more affirmatively worded that we call for.

DR. TUCKSON: So therefore somewhere we call for it, that's the deal?

MS. BERRY: Right.

DR. TUCKSON: Somewhere. It may be at the very end, we are calling for somebody to do something. I guess that's where we're not sure yet.

Ed?

DR. McCABE: One could just restate that first sentence, "We recommend an analysis to determine which health providers." If you want, because we've lost the IOM piece, you could put in parenthesis, "We recommend an analysis, e.g., IOM, HRSA, et cetera," since we know that HRSA has been doing this, and we may be able to utilize that information. But also perhaps trying to recommend that this analysis be elevated to the IOM level, which would recognize the importance of it to health care in the United States.

DR. TUCKSON: I think that's good. I wish I knew more about, and I wish we all knew more about exactly where is the leverage point. I mean, would it be wonderful if it was as simple as if we then sort of sent the letters to the three organizations who have the most opportunity to come together and nail this thing? Maybe CMS would convene it or something, and we could actually call for that level of specificity.

DR. McCABE: Well, I would remind you, Reed, that in the past, one of the things you have recommended is a czar or a czarina of genetics. Again, that is an issue that I think we have discussed, that the genetics services is really quite fragmented within HHS, let

alone the rest of the government.

DR. TUCKSON: What about the committee? The health professions committee at your place, Alan? You guys co-chaired, I think. There were about 30 or 40 organizations that came together. NCHPEG. Is this anything that NCHPEG could deal with? I'm sure it is, but I'm just trying to check.

DR. GUTTMACHER: NCHPEG really is focused on health professional education in genetics. It is trying to be a big tent and invite everyone in who might be interested. I suspect that part of what we're talking about might be to throw some people out of a tent, and therefore, it might be contrary to NCHPEG's very efforts and in some ways compromise its ultimate achievements.

DR. TUCKSON: Well, the IOM, we threw that one out, right? Because first of all, it takes a while.

MR. MARGUS: Because it would be a meta-analysis, Francis said, at a very big picture level only using analyses that other people had already done and drawing them together. So if they exist, we could use IOM. All we needed to do was to do homework to see if it exists, and if they don't exist, recommending IOM would be kind of a ? -

DR. TUCKSON: Well, let me just maybe do this. I don't know whether, Madam Chairperson, on this if we approve everything else to this point, and then maybe actively solicit from the genetic counseling community, and we also have you as chair or someone else, and we just start identifying potential places to get this study done, and then come back with some kind of a recommendation.

I think what we are hearing is we can't solve this one at the table, but that this is a priority low hanging fruit that we want to knock down between now and the next meeting.

MS. BERRY: Ed?

DR. McCABE: The reason why I put IOM back in, I think HRSA is already doing a study. IOM would be a meta-analysis, but it would probably be more back to that 35,000 to 50,000-foot level that we were talking about. What the IOM would do, I think, would identify gaps.

The other thing, the other reason to put it in is a specific recommendation that the IOM doesn't have funding for this. Funding would have to come from someplace. That's the reason to put it in a recommendation to the Secretary, so that there might be a consideration for funding of such a study.

I don't think we're saying that this would be one study necessarily. There might be a variety of studies looking at different levels of the issue.

MS. BERRY: Debra?

DR. LEONARD: Right now, those three recommendations are fairly broad, calling for studies. Yet we have heard from a number of people on the committee, Kimberly and others, who say that the current genetic counseling is useful. There is not enough of it, and people don't get paid.

This is, as Emily pointed out, a coverage and reimbursement document. I don't feel like we're addressing the elephant in the room, which is that genetic counselors can't be paid.

MS. BERRY: The problem is, though, just to play devil's advocate, and I agree with you, that in order to get them paid, particularly under Medicare, a case needs to be

made. We just can't show up at CMS or at Congress and say, well, we really like these people, they do great work, they should be paid.

They will say, show us why. If the evidence exists out there, the data exists and has been analyzed and collected that demonstrate the value, the effectiveness, the importance of reimbursing for these services, it shall be done. But I don't know that we have that yet. I guess that is what these recommendations are aimed at, at making sure that we've got the information that CMS would require, and that Congress would require. They won't just take our word for it.

DR. LEONARD: But there are at least two states that are going to be licensing genetic counselors. There must be a body of evidence that says that licensure is reasonable. Where is that body of evidence that got those two states to provide licensure? It must exist.

MS. BERRY: But licensure isn't necessarily a guarantee of reimbursement.

DR. LEONARD: True, but it is necessary for reimbursement.

DR. TUCKSON: Again, I think we have some work to do. We need to bring that stuff forward right away.

So I guess, Debra, the question for you would be ? - well, can we go back, Sarah? Remind us on this report where we are in terms of when we want this to hit the streets. Is this supposed to be locked and loaded by the end of this meeting?

MS. CARR: No. The plan was to go out for public comment. I mean, I guess there was some discussion we needed to have about when the committee wants to be able to finalize the report. Is February even possible, or is it going to take until June? Is that what you're asking?

DR. TUCKSON: Yes. So there's no question. So just to be real clear so that we're all on the same page, obviously we don't want this report to take 99 years. Does everything have to be solid, tight, and really terrific before the whole thing goes out? Or can you reference that more work may come on certain parts of it? Specifically, on this one given that we've got some evidence to uncover, a little bit more work to do on this topic, would you be willing to get that work done and see it in the full report, or reference it as stuff coming after the report is released?

How important is this issue to this report, and having this one sort of nailed down with a little greater specificity? The enemy of the good is the perfect. Do you want this particular issue perfect?

DR. LEONARD: I don't think we're talking about perfect. I think we're talking about two philosophical differences. One is to call for a study. I would argue that that information exists. We haven't just gotten it yet.

We could potentially in between ask for an update from Judith Cooksey on the HRSA study and the status of that, look at what has been presented for licensure, you know, and I'm sure genetic counseling ? - Peter was just saying that Andrew Faucett is speaking to us in the public comment section this afternoon, and maybe he's here and could provide information to staff on what information exists.

Between now and the next time we have this discussion, maybe we do need to just say, we need studies. But I would think that if two states are providing licensure, that information is out there. We could get it, and we could then make more specific recommendations the next time.

DR. TUCKSON: Well, explicit in your comment, if I understand it, is that it appreciates that this report will come back for another discussion anyway at the next meeting, and that there is another meeting where we will discuss this. We will all see this again as a full committee. In the interim, certain work can occur.

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

MR. GRAY: No, my comment was just simply to point out that Andrew Faucett was listed as a public speaker so that he may be able to address this issue.

MS. BERRY: Emily?

DR. WINN-DEEN: So one other suggestion might be to invite people who were involved in the creation of the licensure programs in, I think it is California and Utah, to come and give us their input. What did they do? What were the studies they used? What were their criteria? What were their goals? Are they already working towards this on a state level? Is this something that we could leverage or specifically recommend the California pilot program be used as the demonstration project?

So rather than just calling for a demonstration project, we might be able to me more specific if we knew more about what was going on at the state level. I mean, to Debra's point, it seems like those states must be pushing towards this for a reason. Not just to have licensed professionals, but probably to deal with the issue that we have before us of how to not just get them licensed and credentialed, but also to get them paid for the services that they render.

MS. BERRY: Not to put you on the spot, but Mr. Faucett, I don't know if you wanted to make any comments now to help inform this discussion, or would you like to wait until the public comment period? What is your preference? We certainly could benefit from your insight on this.

MR. FAUCETT: I'm willing to do either one. Whatever helps the committee the most. Just a quick comment.

DR. TUCKSON: If you could just introduce yourself. By the way, I think this is a good time.

MR. FAUCETT: I'm Andy Faucett. I'm a board-certified genetic counselor, and I'm here today representing the American Board of Genetic Counseling, which currently accredits training programs and credentials about 1,400, 1,500 genetic counselors, the largest group in the country.

In reference to licensure, there are actually three states that currently have licensure, Illinois was recently added to the list. I believe there are 14 or 15 currently working on that process.

It is important to separate the distinction between licensure and billing, and reimbursement. One of the things we all learned is that licensure is to protect the public. It does open the door to billing and reimbursement, but the two should not be directly connected. But I'm here as a resource to answer any questions the committee has.

Yes, Barbara?

MS. HARRISON: I guess just specifically on the literature that is out there showing the efficacy of genetic counseling.

MR. FAUCETT: I'm not sure there is a lot. I know there is some, and I know it is pretty powerful that is there. It might be worth pulling together.

DR. TUCKSON: Do you have any sense of the similarity and/or

differences between the criteria that are used by the three states? Is this very homogeneic, or is it state by state?

MR. FAUCETT: All of the states currently are using ABGC credentialing as their process. Some also have to have another door in, because currently to get ABGC certified, you have to be trained in an ABGC accredited program. So some states have to have another door for people to come in, and also a method for people who chose not to get accredited years ago when it was optional, to consider doing that.

The ABGC actually made a statement at a board meeting about a year ago that we would work with any state to provide a program that met their needs. But currently all of the states are using ABGC certification.

DR. TUCKSON: And the scope of practice also defined, and is that similar state to state so far?

MR. FAUCETT: Yes, it is. I think the only difference is California has a provision about physician supervision, and the other two states do not. I think that is one issue that I think you'll see variation from state to state.

The ABGC accreditation and credentialing process is a competency-based process. So you can go to the website and you can see the competencies that both counselors are trained under, and then the competencies that they are expected to be able to show when they take an exam, or practice as a genetic counselor.

MS. BERRY: Barbara, and then Hunt.

MS. HARRISON: I didn't know if Kelly Ormond from NSGC may have more information about this.

MS. ORMOND: Thanks, Barbara.

Having been one of the people involved in the Illinois licensure, I actually was pretty involved in trying to come up with some of that documentation. I will agree that there is not much of it out there. I think that's very important to recognize. So I strongly support this committee's idea of trying to pull together what literature is out there in a comprehensive manner, because that will be enormously helpful for states going through this process.

One of the things that we did cite in our process was the literature that Barbara was referencing, which looked at outcomes comparing non-genetics physicians and genetic counselors, particularly in the prenatal environment. We looked at termination rates and other outcomes in terms of health, morbidity, and mortality.

But there is almost nothing about effectiveness or usefulness.

I think that one of the things that NSGC has done recently is we put out an RFP that is almost complete that is looking at developing a prenatal model for genetic counseling, and where you can have impact in trying to develop those sorts of models. We would strongly encourage more studies in that area.

MS. BERRY: Hunt, and then Muin.

DR. WILLARD: I think we have to be a little bit careful, especially in this conversation, because the issue, and someone correct me if I'm wrong, the issue is not is genetic counseling effective. The issue is is there a difference between supervised genetic counseling in which you can bill under the name of a physician, and unsupervised genetic counseling in which a counselor might be able to bill on his or her own behalf.

So nowhere is the issue of effectiveness coming onto the table here. So

either we assume as Muin implicitly did earlier, which is that of course it is important and effective, that is why we all want to do it. The issue is simply do you need to have a physician sign off on it and bill under his or her name, or can a genetic counselor. So am I incorrect in this?

MR. FAUCETT: I would argue that you are, because there are many institutions that don't feel you can bill under a physician. There are many institutions who don't bill for genetic services just for that reason. They provide the counseling services, but they don't bill for them, because the institution doesn't believe they can bill for them under a physician. So it is not as simple as whether or not it is under a physician's supervision.

DR. WILLARD: But that's an interpretation by ? - I mean, when I was running one of these services, we went through exactly that discussion, whether we wanted to charge or not under an interpretation of that. But that's not Congress' particular concern.

MR. FAUCETT: I would say that's the largest interpretation currently.

MS. HARRISON: And I think it is partly up to Congress, because it is my understanding the reason why genetic counselors, and not all genetic counselors, it is not acceptable for us to even bill under physicians, because we're not listed under the CMS allied health, I forgot the wording of it, as recognized professions. So that's one of the issues.

DR. WILLARD: Right, but in the context of this report, it does seem reasonable for one of the gaps that we might identify to say that there is a range of interpretations, and the bar that one has to address and get over as an institution to say, do I want to have a physician who is in the same suite who is literally in what does it mean to be supervising, and is that frankly worth the dollars lost to provide such supervision, versus the dollars you might gain by actually getting reimbursed for it. So it is relevant for this committee to address that particular issue. That is a real life issue that every institution goes through, or every genetic service goes through in trying to meet its bottom line.

The point I was making is that it is not simply a question of is genetic counseling effective. Because I don't think that is either in question, or particularly relevant to the issue of reimbursement.

MS. BERRY: Muin?

DR. KHOURY: I'm not sure how to tackle this. We are dealing with reimbursement issues here. We are dealing with the elephants in the middle of the room. I heard that there is not enough counselors out there that can provide services to people that need them with genetic diseases.

On the other hand, those that do provide the services are not reimbursed for them. So we have a Catch-22 here. If we were to commission the IOM report to examine these issues, they would go off in a corner, study this issue for two years, gather data and information, and then write a big report on it.

It would seem to me that this committee would be served by commissioning a paper by somebody, or maybe a smaller subset of the committee, or getting somebody from the outside to write a review of the available literature and here I might kind of defer a little bit you, Hunt, on the ? - I mean, there are many nuances around effectiveness and utility of genetic services and counseling. You can define that any way you want.

We all happen to think it is wonderful and useful. So there is an inherent bias there. If we can through systematic review of the literature, putting all that stuff together with well defined outcomes, I think, and I heard from Andy that maybe there is not enough of

that being put together.

I think this committee can go a long way to address the issues of coverage and reimbursement for these services by beginning to pull that literature together and identifying the gaps.

Ed, you said earlier that it is completely ethical to do a study in this area. I never thought that it would be ethical to have 50 people with the same disease, and then put them into two groups, 25 people that you don't provide genetic counseling for, you just give them a diagnosis, or you can have different nuances like supervised versus unsupervised.

I mean, there are many issues that we can talk about. If the fundamental tenant that these services are useful in some fashion in terms of outcomes, even psychological outcomes, it doesn't have to be completely health outcomes, then why aren't they reimbursed?

I think this committee, we are playing the chicken and the egg here. We are in a Catch-22. Somebody has to take that on. If the IOM takes it on, that's fine. But I think we can do a bit of more homework for the IOM by pulling that literature together ourselves, or commissioning somebody to do it.

MS. BERRY: Ed?

DR. McCABE: I'd just like to clarify what I said. I wasn't indicating that the placebo would be no counseling. What I was saying was if we could do controlled clinical trials in an operating room where they are comparing different approaches, we could certainly look at different approaches and identify what is the best approach here.

So I think it was more that it has been unheard of to do clinical trials in surgery, because every surgery was different. But people are beginning to do those now.

MS. BERRY: Reed?

DR. TUCKSON: I think obviously we need to move this forward. I think Muin's recommendation is one I think that gets at what we need to do. I would speak in support of it.

As I understand it, what it accomplishes is first it allows us to have the recommendation that's on the board for now. What it allows us to do is to push that recommendation forward with a definitive concrete action that we can sketch out the actual paper, the study, the accumulation of data, and the analysis that we look for. We can do that little bit of detail offline.

It allows us to have something very definitive prepared for us by the time we sit down again. Thereby, that puts an urgency towards moving our agenda forward. It gets us off the dime today, because we can't solve this problem today, because we just don't know enough to be able to resolve it.

DR. LEONARD: Would you two be willing to work together to provide and work with staff to provide as much of this literature in some sort of summation form with the papers ? -

MS. ORMOND: Absolutely.

DR. LEONARD: ? - to this committee by some point? It wouldn't be our next meeting. It would probably be slightly before our next meeting.

MR. FAUCETT: Yes.

DR. LEONARD: As well as then I don't know whether Judith Cooksey can provide some information on the HRSA study. But I think those two sources we know are out there and could be used to inform our discussion next time to be able to make more definitive

requests.

DR. TUCKSON: So as I hear Debra's point, it is that the project leader for this, and I think this still fits, Cindy, within your team. Cindy, I think that your team is taking responsibility for pulling that paper together. I think what we've gotten is a commitment on the part of two major stakeholders to be at the forefront of helping us to shape that paper, providing information, guidance, and so forth.

Not exclusively those two, but the others that will probably be added to you. You are truly two linchpins in all of this. I think we all appreciate your willingness to do that. Then we'll let the committee under Cindy's leadership determine others that need to be involved.

Muin Khoury, if you're not on the committee, you are tasked to provide help.

MS. BERRY: Agnes?

MS. MASNY: And I think, too, that maybe what we could also do is ask the International Society of Nurses in Genetics. I know that they were keeping a running list of all the studies and literature that were done by nurses in the field of genetics. So that may also add to this.

MS. BERRY: So for now, in the interest of moving this forward, I propose that we leave these three recommendations as is, with the understanding that we are going to be in the process of selecting information from the folks who are here today, from other organizations, analyzing that, talking to HRSA, finding out the status of their report, and then we may after that be in a position to propose some changes to these recommendations in addition to the substance of the report where these recommendations are found.

DR. TUCKSON: That sounds good to me. Terrific. Thanks to both of you for stepping up to the plate like that on no notice.

MS. BERRY: Moving to the clinical laboratory fee schedule issue, we have heard time and time again from the public and from other stakeholders that oftentimes the costs of providing these tests exceed what Medicare will pay. Remember we are still in the context of the Medicare program.

Lab fees, as we have heard previously, are frozen. So there is the real danger that for the foreseeable future, we will have this gap between what it costs to provide these tests, and what a lab can be reimbursed, thereby providing a distinction for the provision of these services.

This recommendation is not without controversy. The idea was to set up some sort of temporary mechanism for addressing some extreme discrepancies between the cost of delivering the service, the test, and what Medicare will pay. This is the inherent reasonableness capability, I suppose for lack of a better term, that CMS could exercise. But there are some concerns with using this approach. Is anything really temporary?

Some people would view this as a slippery slope. Is this a dangerous course to take? It was the only recommendation that surfaced to address this problem given the fact that we do have a freeze. Payment rates set by law are difficult to change.

So we'd be very interested in hearing from individuals who are most directly affected by this as to what the thinking is on this sort of temporary approach to addressing some of the most extreme cases.

Debra?

DR. LEONARD: I found very interesting the article that was provided to us I believe by AdvaMed. It was by Gregory Raab and Joan Logue. It is astounding to me to look at some of the discrepancies that exist across the board in the laboratory fee schedule as implemented on a state level.

They had some recommendations at the end of the article that they would be more generic than just genetics. If we want to just do the inherent reasonableness for genetic tests, we are talking about 14 CPT codes, billing codes, and the reimbursements for those. This would be a much more limited and directed effort that may be possible to accomplish.

I just don't know whether CMS is willing to look at just those 14 codes in the absence of looking at the entire laboratory fee schedule, which is a disaster. Well, it is not very good, and hasn't been looked at for a very long time. CMS the last time they were here said that it is the oldest fee schedule in existence.

MS. BERRY: Emily?

DR. WINN-DEEN: I wonder if we couldn't be a little bit more firm in our recommendation rather than just sort of asking CMS to look at this again under inherent reasonableness, but to say specifically that we believe all states should be reimbursing at the national level today so that we don't have uneven playing fields among the 50 states where we know that even the national payment schedule is not really covering costs.

I don't know if we have the flexibility as an HHS committee to try and tell states how to implement Medicare programs, but I mean I think that is the first level of inherent unreasonableness that we see when we ask our lab colleagues to come and talk to us about ? - it just seems very capricious and arbitrary.

MS. BERRY: But you're not talking about ? - this isn't the Medicare section. There isn't any deviation, is there, at the state level for Medicare reimbursement?

PARTICIPANT: Oh, yes.

MS. BERRY: But how is it that Medicare wouldn't be directly paying?

MS. GOODWIN: There's a national limit for each CPT code, and then local carriers have the discretion to pay up to that national limit. But certainly it can be lower than the national limit. So that is where the local carrier variation comes in.

MS. BERRY: Okay.

Ed?

DR. McCABE: Actually, from what I know, it is not completely by state, but it is more by region.

MS. BERRY: Region.

DR. McCABE: That's the way the local carriers are. But there is quite a bit of variation from one carrier to another. One carrier may deny services that another finds quite reasonable.

So I think the system, this inherent reasonableness, while it sounds like a bizarre bureaucratic term, is in fact the mechanism for trying to address the lack of uniformity from region to region. So while it sounds bureaucratic, it is bureaucratic, but it is the way the payments work, and the way the approvals are set.

So I think it is a fairly concrete approach to address a problem and need.

MS. BERRY: Debra?

DR. LEONARD: But when you do this process, I don't understand it well enough to know what the impact is at the state and local level. This would be a national

decision. So I don't know what impact that would actually have in practice at the state and local level.

DR. WINN-DEEN: I guess that was my concern was that if even today with a national coverage amount that is not taken up by all regions, you know, and we all know that even if you got reimbursed fully for every test at the national coverage amount, that you are not making any money delivering these services.

So it is a question of how much are the labs losing money every time they deliver a test result? I mean, I think the least we could do is ask the states, regions, or whatever, to step up and be reasonable.

MS. BERRY: Wouldn't it be that CMS would issue a guidance to the carriers that would assist in the implementation of a more fair fee schedule? In other words, the goal would be to eliminate the great variation that would exist between regions, and it would be done through a guidance issues by CMS?

Ed?

DR. McCABE: CMS isn't here, so we're left a little bit in the dark. I don't think CMS is here.

MS. BERRY: Oh, yes.

DR. McCABE: Oh, Dr. Rollins. Yes, please.

DR. ROLLINS: Jim Rollins, Medical Officer, CMS.

DR. McCABE: So could you help us understand what the impact of this would be? My understanding is that it would provide a central guidance. It would still be up to the local carriers as to whether they pursued that. But by giving it some increased visibility at the federal level, there might be an impact by the local carriers. Could you clarify that, please?

DR. ROLLINS: Yes, that is correct. Essentially the local carriers have the discretion to make a decision based on what they feel is reasonable. It is true that there may be some discrepancies comparing one region to another.

To make the request that CMS review the extremes in terms of the variation, I think, is a reasonable request for something for CMS to evaluate.

MS. BERRY: Debra, did you have a comment? I saw your hand go up.

DR. LEONARD: And it is okay to just look at the 14 codes that relate to genetic testing and ignore the rest of the laboratory fee schedule? That's okay?

DR. ROLLINS: I think restricting it to the 14 codes is a reasonable request.

DR. LEONARD: Okay.

MS. BERRY: Any other comments? Ed?

DR. McCABE: Madam Chairperson, can we invite Dr. Rollins to sit at the table so that we can have his counsel in future discussions?

MS. BERRY: Absolutely.

DR. ROLLINS: I'd be glad to. Thank you.

DR. McCABE: I think there is a spot for CMS at the table.

MS. BERRY: If there are no objections, should we close this one out? Leave the recommendation as is? Or are there any changes in the wording that anyone would like to propose before we move onto the next section?

Debra?

DR. LEONARD: I would like to make it a little more forceful and not CMS to assess, to determine whether fees should be changed, but to basically ask CMS to use the

inherent reasonableness to look at the CPT codes and reimbursement that are used in genetic testing, and assure that the current reimbursement level at least covers cost.
That would require input on what it costs to do the testing, but right now it doesn't cover costs.

DR. WINN-DEEN: Can we capture the whole local versus national issue somehow on that, too, to encourage all local providers to come up to the national level?

MS. BERRY: Something to the effect that CMS, or that we're directing CMS and requesting CMS to use inherent reasonableness to examine the variation in payment rates or reimbursement rates for genetic tests and laboratory fees with a view towards ensuring that the reimbursement level in all regions of the country at least cover costs, or something like that.

DR. TUCKSON: I don't know. Just as a minority, I think that we want to be careful. Sometimes I think we want to obviously signal that we have a bias as it were about something. In this case, I think what we ought to be doing is signaling that we have a concern about this, and it needs to be studied in a clear and dispassionate way. You're talking some very complicated issues around money.

For us to conclude in the recommendation that these things are not, you know, that what we want is ? - I mean, this sort of signals the way it says is that we expect them to pay more money. I think we just need to be a little bit careful about being too passionate about that and let them do their work and see where it comes. I may be in the minority there, so I'm drawing that out.

DR. ROLLINS: I think that the request in terms of what is currently on the board is very reasonable. I think that to go beyond that is something that I myself feel that I could not make that commitment. But I do think that that is something that will be studied. If it was felt that there was a marked discrepancy and it needed to be addressed, then that is something that CMS would address in the future.

DR. LEONARD: I guess what I wanted was if there is a discrepancy found, that they fix it. They could do all the studies in the world and say yes, it is broken. And there we would still sit.

DR. ROLLINS: And I would respond and say I hope that corrective measures would be put in place.

MS. BERRY: Hunt?

DR. WILLARD: I just want to make sure that we're making adequately the case prior to this recommendation, that in fact there has been a harm and a foul. So other than what I accept fully, laboratories have a tough time making ends meet, and they are under budgetary pressure.

From the Secretary's standpoint, I'm not sure that is very high on his personal agenda, unless we can argue that because laboratories are under-reimbursed, or if it is found that they are under-reimbursed, that that actually has an impact on health in this country. The tests are therefore not being taken advantage of, or not being provided to members of the public who should be taking advantage of those tests.

Unless we make that case, I'm not sure the simple issue is we want geneticists and pathologists to be able to make the money that they would like to make. I'm not sure that that will resonate terribly well, no matter how well it is stated.

DR. LEONARD: But maybe we could take a 5,000-foot view on this one, too. Which is if we are moving toward genomic medicine and this is going to have such high

penetrance in the entire practice of medicine, then it is a big problem that laboratories don't get even paid what it costs to do the test.

MS. BERRY: Emily?

DR. WINN-DEEN: I think we've heard some testimony before this committee, and we have another commentary in here from LabCorp, which is one of the largest reference labs in the country that it is a problem, and that the choice they have is to bill the patient for the difference. I don't think it is too big a leap of faith to take that jump that some patients won't be able to pay that.

DR. WILLARD: I personally don't disagree. I'm just suggesting the committee needs to connect those dots in the report so it doesn't seem like just a request for more money for those providing the test. That's all.

MS. BERRY: We can perhaps take a look again at the language and the substance of the report just to make sure that it forcefully enough states the case that you are articulating. We're not just calling for change in reimbursement just for giggles. I mean, there is a compelling need there related to access, and it falls within the reimbursement charge that we have in doing this report. We'll take a look at that and beef it up if necessary. That will be presented to everyone once again.

Just to tie this up, we floated some language, but it sounded like Dr. Rollins was a little uncomfortable with the revised version. Should we go back to the recommendation as currently stated? Or are there some tweaks that would state what we need to accomplish, and CMS would nevertheless be comfortable with it?

We don't want to recommend something that is not going to be implemented. That will produce no benefit at all.

Ed?

DR. McCABE: Well, I think we heard that CMS would be most comfortable with the language as it is on the board. I think we could put the rhetoric into the text. Not of the recommendation, but of the logic that concern has been expressed to this committee regarding the ability for the laboratory to mute its expenses, or something to that respect. So I would put the rhetoric in the report and leave the recommendation for discussion.

MS. BERRY: Any objections?

(No response.)

MS. BERRY: All right. Let's move on to Medicaid and SCHIP.

DR. LEONARD: Could we go back one second? You may just want to say in there so that you are defining the ? - to look at the specific CPT code. The ones that do exist, so you're not talking about ? - well, maybe that's implied by genetic test laboratory fees. But it is a limited scope that we're asking them to look at.

DR. WINN-DEEN: So you just want to add the 10 or whatever?

DR. LEONARD: Existing CPT codes, or whatever. Never mind.

MS. BERRY: We'll fix that.

Medicaid and SCHIP barriers. Of course there is a great deal of variety from state to state, because each state has its own programs with regard to Medicaid and children's health insurance.

There are some reports that we have heard about with regard to instability and coverage for genetic services. States are having difficulty balancing their budgets. So we had two recommendations to propose for the committee's consideration.

One would be really an information dissemination function for CMS that states could benefit by HHS providing the states with information, the best information, and establishing the solid foundation and evidence base for covering and providing genetic services.

The idea being that if presented with this information that they may or may not already have, that the states would be more likely to cover these services in the programs that they implement. The second recommendation is a little bit more of a carrot which would be to provide states with actual incentives, presumably financial incentives, to cover genetic services that are warranted by the evidence base.

That is a little bit more difficult because it involves money that may not be there. But those were the two potential recommendations for encouraging states to provide these services, and covering these services. Recognizing that we really aren't in a position to mandate that these services be covered.

Does anyone have any comments on these potential recommendations? Like them? Hate them? Revisions? Debra?

DR. LEONARD: Do we have an idea of where this evidence base is coming from? I mean, maybe we should put e.g., EGAPP, or other HHS initiatives that may inform what is going to be provided to states as examples. Maybe HHS representatives could inform us about which programs to put there as examples that could inform this.

MS. BERRY: Barbara?

MS. HARRISON: And then to add to that, whatever we're able to uncover about genetic counseling specifically. That tends to be a great challenge with Medicare, or Medicaid, rather.

DR. TUCKSON: I guess I'm a little confused on this one in terms of this incentives business. I mean, we've got a $50 katrillion deficit, and HHS, I mean, boy, I'm just trying to figure out what would be the financial incentives that they would ? - how would they think through that? I mean, what do you do when you get this recommendation?

If you just have a sound evidence base, is it a priority? I mean, do you provide the incentive for this and not the first trimester prenatal care clinic in Delta Mississippi? I'm just not sure, how are you asking them to think this through?

I mean, there is no new money, so they are going to take it from someplace else. So how do they think about this?

MS. BERRY: Again, I'm going to throw this out there, because this has worked in the past. My own view is it is not a realistic recommendation in this current budget environment. We have put them all out there for everyone's consideration.

We could, using precedent, recommend that there be some sort of a grant program. I mean, I know that the Secretary has issued grants for information technology, for example, unrelated to this, of course. But there can be state and local communities that would benefit from a grant that would provide an incentive to offer these services. Again, we would have to answer your question, which is right on, of course, Reed, which is does this rise to that priority level, given all the other services and needs that are out there.

Emily?

DR. WINN-DEEN: I guess I have a little problem with recommending grants when we don't even have a standardized newborn screening program which we know works, and we know there is a lot of evidence that that pays itself back. We as a country have not been successful in taking some kind of a national minimum standard approach and

disseminating that out.

Right now we are being embarrassed by the March of Dimes into trying to do that. But they are really leading the charge, the federal government. So I would feel comfortable with us saying that, you know, when stuff gets to a certain level of evidence, whatever that is, that then that information should be disseminated to the states, and the recommendation from the federal level should be that all state programs adopt that.

Whether they are able to adopt it immediately or they have to think about what the tradeoffs are within their budget, you know, I don't think we can really propose anything more than guidance to the states.

Maybe Suzanne, do you have more information? Are there any grants for newborn screening or any other kind of underwriting for basic programs like that?

DR. FEETHAM: I have a summary of that from MCHB for '05, and what they have done in '04. Also, Reed Tuckson is our representative from this group on the Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. That report is in process now.

It is my understanding that it specifically will advise the Secretary regarding the universal newborn screening technology tests and programs. So we may have more to say about that.

DR. TUCKSON: Actually, the good news here is that I have been mercifully fired from that committee, and better representation has been found. So the world is a better place as a result. In fact, who is our person? It is important, actually. You need to know that we've upgraded our strength on that.

MS. CARR: Actually, we haven't asked him yet.

DR. TUCKSON: Oh.

MS. CARR: Maybe we should not say.

DR. TUCKSON: I see.

MS. CARR: It's in process.

DR. TUCKSON: Well, in process and he who it is that will be doing this, even though he doesn't know it yet, we thank him for it. He is much better than I am.

The point that I think we have on the table ? - thank you, Sarah. Ask him at the break so we can announce it. He has already agreed. I think with the issues that are on the table are, if I understand it, first is we've got enormous state variability in terms of various mandates for programs, and it drives anyone nuts. The evidence is there, or it isn't there. You've got all this up and down all over the states.

We have clearly got here the idea that there is as a priority area of genetics, particularly since we put SCHIP there, which means the children, is that you've got the Newborn Screening Act, which is all over the place. As you said, what you'll hear, and we have an official liaison to that committee, is that you'll see that every state has ? - it's all a complete mishmash of stuff in terms of what kids are getting access to or not. So they are trying to rationale and streamline that out. That might be a good place to sort of start.

But I think the final thing is that for at least as a perspective for our committee, maybe one of the things we ought to be calling for providing is some sense of a common evidence basis that can inform the decisions that people make, as opposed to saying you ought to provide incentives for covering things that have an evidence basis.

That there are decisions that people have to make based on their priorities. I

think the only way this will make sense to the reader is if we give some tangible examples of the kinds of things we're talking about here.

DR. FEETHAM: Also, to add to that, MCHB provided funding last year for regional newborn screening and genetic collaboratives. There were seven regions funded with the National Coordinating Center located through the American College of Medical Genetics.

So again, it is recognition of the issues that you're saying that this funding is in order to try to address the issues you're talking about of the disparate distribution of the services. Again, that is just responding to the fact that MCHB is funding these types of efforts.

MS. BERRY: Ed?

DR. McCABE: And NICHD has recently begun an initiative in newborn screening and has recruited former chair of pediatrics at the University of Miami Rod Howell to take that on. So I think this is a broadening. It is a recognition within HHS of the importance of newborn screening.

With two agencies taking an active ? - well, actually three, because CDC has for a long time had quality assurance activities there, and there may be others who I'm not mentioning. But I think there is a broadening recognition that newborn screening as we've heard before, will be the leading genetic testing for the next decade. Most likely with 4 million babies born every year, and every baby having somewhere between 4 and 30+ tests, this is a huge genetic testing undertaken. It certainly needs to be standardized.

DR. WINN-DEEN: Do we want to make a specific comment that we support current efforts underway to achieve a standardized level or nationalized level of newborn screening as a good starting point for that?

DR. McCABE: Not to get into turf issues, but I think it is a bit of a turf issue. There is another Secretary's Advisory Committee. I think that we probably ? - I would think it would be more appropriate for that group to report to this committee and after such report, then go forward with the recommendation. I think it would be acting on hearsay at this point not to have a formal relationship and a formal report.

MS. CARR: Actually, during the last session of the day, we were going to consider other topics that could come up in February. Chris Hook, who is not here today, but very interested in newborn screening, had actually suggested that we have a presentation from that committee about their recommendations. So we're going to talk about that later today. Or we can decide now if you'd rather, to for sure have that in February.

MS. BERRY: Muin?

DR. KHOURY: If we're talking about disseminating evidence base for genetic testing services in general, maybe I missed part of the conversation. I think there are ongoing efforts other than the newborn screening area that are going on with states.

The CDC has funded four states in chronic disease programs to begin to take a look at these things, sort of what you are alluding to here, trying to figure out how genetic testing and services can be used outside the scope of the traditional MCH arena in genetics. We have funded schools of public health to begin to build that evidence base and provide technical assistance to state chronic disease programs. So that is sort of another thing in the hopper.

MS. BERRY: Would everyone be comfortable if we kept the first recommendation subject to any revisions or tweaking that anyone might want to propose, but

eliminate Number 2? I have sort of heard mixed things.

One is that, you know, the budget reality is such that it would be difficult for us to propose or for the Secretary to offer actual dollars to states. On the other hand, there are grant programs that currently exist, and we don't want to impede that progress.

I don't think an omission of Number 2 would necessarily have any adverse impact on the existing grant programs. But what are people thinking in that regard?

Emily?

DR. WINN-DEEN: Well, I think maybe what we need to do is just change the words from "incentive payment," which is in my mind like a little carrot that you're holding out to actual specific grants, which is I think what is actually happening to provide demonstration projects and to assist in getting some of these things implemented. I think that might be more reflective of the actual practice within HHS, which is already happening. There is obviously some funding set aside to do that, so I would be okay with that.

DR. LEONARD: And it could be worded that HHS continue to provide states with support or grants, indicating that they are doing that by various mechanisms and continue to do that to implement the sound evidence base of testing.

We've narrowed this down to Medicaid and CHIP. But if you look at adults, we want genetics to be used on an evidence-based mechanism for all of genetics. Do we want to broaden this potentially? Would there be any utility?

Muin, I know you're looking down, so I can tell you're working on your blackberry, so I'll get your attention first.

DR. KHOURY: I know how to multitask.

(Laughter.)

DR. LEONARD: Yes, but this wasn't one of the tasks.

(Laughter.)

DR. LEONARD: So is it useful making recommendations at the state level for Medicare? Maybe this is more CMS, but you are the one that is generating those evidence bases. So could we broaden this to be Medicare/Medicaid/CHIP coverage decisions? Because I don't know how much Medicare is at the national level versus also being influenced at the state and local levels.

MS. BERRY: Well, later on in the report as well, just to remind everyone, there is the section that deals with all payers. So there are some recommendations that we're going to be working on which address and get to the evidence base, that get to how do we make these decisions? Who covers what under what circumstances? So I don't know if that is a good place to get at your point.

DR. LEONARD: Maybe something once we work through all these recommendations -- I'm getting the feeling that they're not lumped. This is the process you have to go through is you take the body of what you have written and you kind of duplicate that in creating recommendations.

But like the UPIN for genetic counselors could go with the genetic counseling if there is stuff on Medicare and private insurers, maybe this dissemination could be lumped all into one recommendation so that we don't end up with 50 different recommendations if we can lump them and make fewer, it might have greater impact. Even if it is down the road after we work through absolutely all of these individually.

MS. BERRY: It is quite possible we may have to ? - well, we are already

reorganizing the paper from yesterday's discussion. So we will definitely need to do that after we get through these.

Muin, I sort of intervened.

DR. KHOURY: No.

MS. BERRY: So any final thoughts? So what I heard then most recently was we keep Number 1. Number 2 we revised slightly to reference the fact that HHS is already providing some grants or some assistance to states, and we would urge them to continue to do so. That gets us away from HHS must set up some new program and provide actual dollars to people, but more it is an encouragement of existing policy, and to the extent possible and feasible, they could expand that within their discretion.

Any other comments?

(No response.)

MS. BERRY: All right. Now we're in the ? -

DR. TUCKSON: Cindy, we are at our break time, actually. Would you mind if we stopped here for a minute?

MS. BERRY: Not at all.

DR. TUCKSON: Terrific. You're pushing us through.

By the way, let's just do a process check again. We are at our 10:15 break. We then have from 10:30, which we'll more back up a little bit, to 12:30 to conclude this section. We have about 15 pages left, so I think we're actually on time. We'll probably get right to where we need to be. I'm not so concerned.

You're doing a great job, Cindy.

Why don't we reconvene at 10:40.

(Recess.)

DR. TUCKSON: Cindy, take it away.

MS. BERRY: All right. We are now in the section where we are addressing barriers that apply to both public and private insurers. The first section in this part of the report deals with the fact that Medicare is often a model in many other circumstances for private insurers.

So to the extent that Medicare has coverage and reimbursement problems with regard to genetics tests and genetics services, it could have a ripple effect and an adverse effect on coverage and reimbursement in the private sector.

However, as a task force, and as a committee in the past, we've struggled a little bit with this because while that may be true in many other fields, it does seem, because we had heard some testimony and received comments from folks in the private sector, that in the area of genetics, many times the private insurers are a little bit more advanced than Medicare, because they don't have some of the same legislative constraints that shackle CMS in terms of what can and can't be covered. So, for example, the screening exclusion that doesn't really exist in many private health plans.

We recognize that Medicare is often a model, and so that is one additional reason for changing or facilitating changes in policies that would expand access coverage and reimbursement for genetic technologies. We struggled a bit with what the recommendations should be.

This isn't really in the form of a recommendation. It is more of a statement which basically says that private insurers should not wait for Medicare to make these

determinations, and that they should essentially without directly saying it, make their own decisions based on the evidence base.

This may not be worthy of a recommendation, because it is sort of a declaratory statement. We talked also about possibly advocating for some outside organization, whether it is AHIP or some other entity, to develop voluntary guidelines, or some standards that would not be mandatory, but that would be a model for other private insurers to adopt with regard to genetic tests and services.

We didn't put that up there. This is one that we struggled with, and we could certainly benefit from the committee's views on.

Ed?

DR. McCABE: I think it's quite good. I think it is important for it to be there. It is a declarative statement, but that is what a recommendation is.

I would just make a couple of minor wordsmithing things. The last line in the book at least, let me just read the second sentence. "Such services should be considered specifically with respect to the benefits." Let's be positive. To the benefits, they offer the populations they serve. I'd get rid of "different" also. I mean, I just think we waffle what they can offer. If there are benefits, then they should provide those benefits to their clients.

MS. BERRY: Hunt?

DR. WILLARD: I would disagree with Ed, only from the standpoint that we're providing recommendations to the Secretary. So having it in the text as a discussion to provide background is fine, but to make a recommendation to the Secretary that others who are well beyond his reach should be allowed to do what they want to do seems like an odd thing to be doing.

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

MS. MASNY: I was just going to comment that since we have been talking so much about evidence-based practice, that many of the health insurers now have their own technology evaluation committees, that maybe in the text, as Hunt was just saying, we could say that with their own appropriate evidence-based review, that genetic tests when appropriate, that they don't have to wait for Medicare. They could move ahead with the testing.

MS. BERRY: Now, I will point out that later on in the report, and we'll get to that, there is a section -- and in fact we're going to restructure it even more than what currently exists in front of you -- there is a section on information utility, the preventive nature of genetic services, factoring costs into coverage decisions. There is another thing that we are talking about moving.

It all leads to the evidence-based discussion. The recommendation there, without jumping too far ahead, deals with HHS establishing some sort of working group to come up with a set of principles that would help guide insurers, public or private, in determining what should be covered, when they should be covered, and when certain services and tests should be covered, that kind of a thing.

So I don't know if that could serve as a recommendation for this scenario. Maybe we are lumping instead of three or four, it is all five of the barriers that have been identified, this being one of them, and have that recommendation serve as the recommendation for all four or five.

Ed?

DR. McCABE: I understand the reason for removing it as a

recommendation to the Secretary. I have no objection to that. But in the text, let's make it clear that we recommend that private insurers not wait for Medicare. So let's make it clear, because I don't want to give the private insurers an out that until Medicare is ready, we're not ready.

If we do that, then we need to also look at the other recommendations within this section, if we're going to go by that principle. But we can state them as recommendations, highlight them in the text somehow, but not as a recommendation to the Secretary.

MS. BERRY: Suzanne, and Reed.

MS. GOODWIN: I think to get to Hunt's point, and also Ed's, certainly the direct audience that this committee has is to the Secretary. But certainly I would like to remind you that you also have a national audience. While you might not have as much weight in addressing it, this report is going to be available to the public.

I would hope, especially in addressing the various barriers in this section, you would keep that broad audience in mind and not feel limited in any way to addressing your recommendations to the Secretary. I hope that provides some clarification to you as you work through some of the issues. Certainly this topic in particular has broader relevance than just Medicare. I hope that you don't just feel limited to crafting any recommendations directly to areas that the Secretary has control over.

Reed?

DR. TUCKSON: Yes, two things. I think first that last point, that last dialogue is important. I continue to come back to the idea that the committee will be best served by focusing as much as possible on its priorities. If we're just scattershot all over the place, we are not going to be as effective as we would be if we focused in on things that are the most important.

Having said that, I think that the recognition that there is a broader audience than the Secretary that we do speak to is important. I think we shouldn't speak only to those things which the Secretary ? - maybe we might want to bring it to the attention of others, or in the course of our deliberations, we have uncovered this important issue.

If it is important enough, we may want to speak to it. The only issue I have with this recommendation here is that the private sector, as the text says, is doing these things. I think we want to be careful that if we're going to make a recommendation that it doesn't look insulting.

You know, the way it appears now is as if the private sector was simply not doing anything and waiting for Medicare to act. That is not what the text says. So I think you want to sort of maybe encourage them and so forth.

The other thing is if we're going to add the insurers on this one, it is also the payers. The way in which we set this up in the earlier preambles, it is the combination of how you get to these things is ultimately determined by the purchaser, and the plan. So it is all of those folks working together.

I think the final point is just really grounding this stuff. Again, evidence basis, and priorities.

MS. BERRY: Does anyone have any suggestions as to whether this should stay as a stand-alone piece with this recommendation as revised by some of the comments we just heard, or whether it should in fact, and perhaps we can't make that determination until we get to it, but lump it in with the other section which has a recommendation dealing with

evidence base? Does this merit really standing alone in isolation because of the critical importance of a statement that we want to make here? Or can this be merged in with the other items? Does anyone have any strong opinion?

Debra?

DR. LEONARD: I think given that private insurers are to some extent at least doing this, I don't know that we need to stand alone. I think by incorporating this into the evidence-based section, we can still make the same impact and have fewer recommendations overall.

MS. BERRY: I think that's a good approach. Does anyone object to that? So we would remove this recommendation, but merge it in with the other section dealing with evidence-based practice. We would still retain the text, it just would be in a different section. Hearing no objection, that's what we'll do on that section.

We also had discussed very briefly the UPIN issue, and discussed moving it into the section that we addressed earlier, genetic services and counseling. So it wouldn't be a stand-alone here, it would move into that section. So we can still go into the recommendation, but just know that it will be moved into the earlier section that we talked about.

This is an issue of course that identifies the fact that genetic counselors are not eligible for a UPIN, and that many health plans use the UPIN system. So if you're not eligible for a UPIN and the health plans use that system, that may adversely affect the ability of genetic counselors to directly bill private insurers.

The recommendation would be until the national provider identifier system is implemented, that perhaps private health plans could create their own provider numbers for genetic counselors to use for billing purposes. This also falls into a similar type of situation as the earlier recommendation.

Well, it is a little bit more of a recommendation than a declaratory statement. But I think health plans do know that they are at liberty to set up their own numbers if they wish. The question is whether the committee feels that it is important to make that statement, and urge them to do so.

Ed?

DR. McCABE: I would make the statement, but I'd include it in that string, I think it would be Number 4 then, if I recall, 3 or 4 in that string that we had that we started the morning off with.

MS. BERRY: Debra?

DR. LEONARD: I would say that this potentially could be informed by whatever information is brought back to the committee at the next meeting, and we can move this one up there. But there are certain steps that will have to be taken in order for genetic counselors and those providing genetic counseling services to bill and be reimbursed for those services that are currently going on.

If this is one of the steps that needs to be done, then maybe information could be provided about how that would happen. So including this in that discussion would be useful.

MS. BERRY: Does anyone have any other comments on this?

(No response.)

MS. BERRY: Suzanne, do you have what you need? All right. The next three barriers that have been identified, there is not one recommendation that will come after

each one. You'll see in the report they are lumped together. The recommendation would apply to all three.

It is informational utility and medical effectiveness as one issue. The next one is the preventive nature of genetic services, the third is factoring cost into coverage decisions. Those three things after discussion in the report, they lead to one recommendation which we'll get to in a second. But I wanted to call your attention to that.

This one, the informational utility medical effectiveness piece focuses on the fact that health plans use medical effectiveness to make sure that the services that they cover meet evidence standards. It also in the text discusses the fact that there are some genetic tests and services that people may find useful and informative, but may not warrant coverage because of the medical effectiveness criterion. This raises the issue about whether informational utility on its own really warrants coverage.

The second barrier deals with the preventive nature of genetic services, highlighting the fact that of course there are long-term benefits to providing these services, and they can be cost-effective over the long haul. But in the short term, because people change health plans, that's one reason, coverage for preventive services might be difficult to justify, and insurers may or may not feel that coverage would be warranted.

The third barrier issue that is addressed here in this section deals with the fact that there is some uncertainty about whether and how best to incorporate cost-effectiveness data in coverage decisionmaking. There seems to be a lack of data on cost-effectiveness of genetic tests and services. That has potentially an adverse impact on health plan coverage decisionmaking.

So we came up with a recommendation that potentially could address all three of these issues. That would involve the Secretary tasking an appropriate group or a body establishing a task force so to speak that would develop a set of principles for coverage decisionmaking for genetic tests that would assist employers, and it would assist health plans.

It wouldn't be a mandate, it wouldn't be a requirement, it would be more in the form of guidance. These principles would identify criteria that would help health plans and other payers determine when should genetic tests be covered. When should they always be covered? Which tests should never be covered, at least under the current evidence base. Which genetic tests fall into that gray zone where it might need to be determined on a case-by-case basis?

Cost-effectiveness could be addressed here, as well as the preventive nature of genetic tests and services. Again, emphasizing that this would be more on the lines of guidance. I don't know to what extent some of this work is already being done. We might want to reference that. Does anyone have any suggestions, comments, or thoughts?

Muin?

DR. KHOURY: Maybe you can help us flesh this out a bit more. I thought the set of principles related to genetic tests have been kind of set forth by SACGT earlier, sort of the ACCE paradigm. But it didn't go far enough with respect to reimbursement.

I mean, it basically talked about the issues that need to be considered when a genetic test is being looked at from the analytic validity, all the way to the ELSI issues. What we have taken with the ACCE project and the EGAPP project is tried to move the ball a bit further down the field, if you will.

I think where you end up in a stumbling block is what is that threshold at

the end of the day when you have collected all the information and identified all the gaps? Then the principles around the criteria for what to fund and what not to fund is always a sticky point.

So I'm struggling with a notion here, and maybe others can help by jumping in. Identifying the principles which I think SACGT and you and others have kind of ? - I mean, it is easy enough to say these are general principles, but how to move them forward with the question of threshold. That is a more sticky point. I'll just stop here, and maybe collect my thoughts more.

MS. BERRY: Debra?

DR. LEONARD: I don't understand how this is different than the ACCE and EGAPP. You have to provide information about specific tests, their utility, what they do, when they should be covered, what populations, and I think that is what ACCE and EGAPP are doing.

To me, this sounds like not only you're going to provide that information to Medicaid and SCHIP and Medicare and private insurers, I mean, you basically want wide distribution of that information coming out of ACCE and EGAPP. I think that is what is being asked for here.

I don't know that we can do the cost analysis. I think each provider has to figure out what they can pay for and what they can't. But once it is medically useful, more than likely it will be paid for by some groups, an increasing number of groups over time.

MS. BERRY: Yes, sir? Dr. Rollins?

DR. ROLLINS: I think you may want to consider either removing cost-effectiveness, or putting it at the very end, only because health plans usually don't take into consideration the cost of a technology, at least at the initial assessment.

I think that that would give the impression that cost was being taken into consideration for a plan to consider a particular technology. So either eliminate it, or if you want to keep it, I would stress it as the very last component of that sentence.

MS. BERRY: Barbara?

MS. HARRISON: I just want to make sure I understand. Are groups like EGAPP, which I guess is under ACCE, or follows an ACCE model, are we saying that we don't really need to establish a group then? That there already
is a group established?

MS. BERRY: Well, that's the question. Do we feel that the work that is already being done, is that sufficient? Does that provide sufficient guidance from our standpoint to health plans and pairs? Or does there need to be a new group that kind of synthesizes and brings together all of the work that is currently being done, and perhaps fills in gaps, and then serves as a guide?

Muin?

DR. KHOURY: It seems to me that this committee can make a recommendation to HHS along those lines, because EGAPP is an experiment. Three years from now, it will disappear. Whether this could precede or secede, or be part of EGAPP, or EGAPP could be independent, it doesn't matter.

I think HHS should take the lead, and all the agencies presented here can come together, beginning to take what has been done so far, and then melding it into the set of principles. Then the agencies can figure out how best to work together, or HHS can tell us.

Because we have been struggling with this for a long, long time. I mean, starting with Tony Holtzman's NIH/DOE task force. I feel like every year we make a little bit more progress in this area. Where you get at the end of the day, you get at the stumbling block of people looking at the same data, but making different conclusions with respect to reimbursement and coverage.

Depending on where you set the bar, you want more or less medical effectiveness versus other things. For example, there is a clear distinction between what the U.S. Preventive Services Task Force, and my friend here is not here to defend himself, with respect to the very strict criteria for clinical utility, versus some of the other models of technology assessment, including ACCE that have been proposed.

That is why what we wanted to do at the beginning of the EGAPP project is to put all the methodologies together and try to come up with hopefully a consensus methodology for how these evidence-based reviews can be done. Because again, as I say, it is a step along the way. But if you push HHS to keep its appropriate leadership role in this area with all the agencies contributing to this effort, I think that would be a wonderful goal.

MS. BERRY: Reed?

DR. TUCKSON: Yes, I guess I'm struggling with this one as well. I think maybe Muin got us there. I can't quite tell what we are doing. Let me ask specifically.

Are we taking a position that is an advocacy position that says that at the end of the day, what we want to see achieved is for tests that do not have a therapeutic import, we want to make sure that those tests are covered? We want to see them pushed. Our real issue here is to push in a certain direction for certain kinds of things to get covered.

As a result of that, we are sort of dictating that direction. Or are we saying, this is a complicated world where there are new issues that emerge because you've got diagnostic tests out that are predictive in value that may not be related to therapeutic interventions per se.

As such, it raises the need for information to be readily available for decisionmakers to be able to make appropriate decisions. The categories of information that must be made available are evidence basis of work, you know, the clinical utilities and so forth and so on, cost-effectiveness of what it means to implement this, how this new test relates to existing, if there are any ways of evaluating that disease condition, and so forth.

So what we are ultimately trying to assure is that people who have to make tough decisions have a knowledge base from which to make it. I can't tell whether it is that we view our responsibility as ensuring that a knowledge base exists in a set of areas that are decisionable, or are we saying no, what we want to do is have a knowledge base that allows certain things to actually happen because we want these tests to be implemented today. I can't tell which one we're asking for.

MS. BERRY: Also, I think, and just to mention again, later on in the report, we talk about the evidence-based issue, evidence-based coverage decision. In that section, there are recommendations which include HHS to task a group to assess the evidence for specific tests and determine whether the evidence is sufficient.

You can make an argument that that whole discussion as you outlined the latter point, not your first point out ? - I don't think we're going in the direction of we should cover everything, no matter what it is, whether it leads to some potential therapeutic benefit or not. I don't think we're there, but more the latter point that you raised, Reed, which is there may

be some need to provide guidance in assessing all of these factors may be that we merge this section in with the evidence-based section and take a more global approach.

DR. TUCKSON: And I think maybe the way also, that sort of makes sense to me. What we sort of say is a reasonable, prudent analysis of this new world that we're in means that these new tasks must be considered by any reasonable group of people in the following areas.

Evidence-based, cost-effective, yada, yada. Here is what we now know in terms of the availability of these kinds of analyses. You get to what Muin said in terms of you've got some efforts that are already pulling this together.

However, they are, in our opinion, inadequate, or need to be bolstered or supported by some other new things that people will have to have available to them. What we are saying to the Secretary is you've got a bunch of federal agencies, NIH, FDA, AHRQ, CDC, you've got all these people, and you control access to the kinds of data, or you should be creating the kinds of databases that allow reasonable and prudent people to make intelligent decisions.

We have really got to alert you, sir, that we need this research infrastructure, this information infrastructure, available right now. That means you've got to bolster this. Otherwise, CMS is in trouble, as is everybody else out here in the world.

MS. BERRY: Agnes?

MS. MASNY: I just also wanted to bring up comments that were made at some of our past meetings from the FDA or ex officios from the FDA and the FTC, that if there were guidelines like this, that they had actually requested that these guidelines then could be shared with their respective departments. That would also help provide guidance in their oversight of some of these tests.

MS. BERRY: Yes?

DR. ROLLINS: Something else that Reed had just mentioned, which I just wanted to add to, also. When looking at a screening or a diagnostic test, those tests should be done if based on the results of it, there is something that you can do with it. If there are diagnostic tests available and they might show a certain something being present or absent, but if you can't do anything about it in terms of patient management, which would result in a better outcome, it is probably not a good thing to do.

In adding to what it is that you said a few minutes ago, I think that we should also take into consideration what actionable action is going to result from those diagnostic tests.

DR. TUCKSON: Would you consider ? - and I think the essence of this section as I read the report is would you consider a counseling intervention, Huntington's, would you consider that counseling opportunity a specific intervention that would then make that activity worthy of your analysis?

DR. ROLLINS: Yes, yes.

DR. TUCKSON: Let me just for the sake of argument. Would you consider in utero information about the sex of your baby, you know, or the color of their eyes, would you consider that information worthy of your analysis?

DR. ROLLINS: No.

DR. TUCKSON: Suzanne's asking what makes the distinction, and that's the point. What is the guidance? Or is part of the recommendation here how do we make some

sense of what are the guidances here? Should the American people pick up the tab for everything possible that a person would want to know who is getting public insurance? Or should it be limited to certain things?

I think that's the other half of this whole recommendation. How do you help people to think through? So Part A is you've got the information. Part B is how do you help decisionmakers to think through what is a reasonable use of that information.

MS. BERRY: Debra?

DR. LEONARD: I'm really disturbed by the argument that if there isn't anything you can do, then it is not worthwhile doing the test. I know from a personal perspective, if there is something wrong with me and there can be a diagnostic test that says it is either X or Y, and there is nothing that can be done, I want to know whether it is X or Y.

I want to know what my diagnosis is. I think from a physician perspective also, if you have a definitive diagnosis, you stop looking, you don't do other tests. There is utility in diagnosis, even when there is no therapeutic intervention.

DR. TUCKSON: So we go back to the ? - just to be provocative for a minute. What I think makes sense from what I hear is if you knew you had a diagnosis that would affect your reproductive decisionmaking, or at least inform it, that might be useful. If you had information that would affect the way in which you related to environmental toxins or personal health behaviors that you could actually change your health behavior, that might be important.

If you just, again, to know something about the color or sex of your baby, would that be enough reason to know? Or is it somewhere in between?

DR. LEONARD: I don't think color of eyes or sex of an infant is considered a disease. So I think the distinction there is that is a personal characteristic, if you will. I mean, you get into gray zones where there are characteristics that are sometimes considered personal characteristics, like obesity and things like that. Although one could argue that obesity is the most prevalent disease in the United States. But I think color of skin, color of eyes, sex of the infant, none of those would be defined as a disease.

MS. BERRY: Emily?

DR. WINN-DEEN: I just wanted to remind people that this is in the section that is not just on public insurance. It is in the section that is on sort of all insurers. So we do need to consider that there is also each insurance company in collaboration with their customer, which is typically an employer buying a benefits package.

They can buy a benefit package that includes some tests, and doesn't. At some point in the future, employers might want to offer, you know, the blue-eyed, brown-eyed gene test for, you know, fetuses. God knows why, but if they chose to offer that as a benefit, as long as it was accurate and valid, they would have that right to do that.

So we should be, you know, there is a difference in what we pay for with public money, and what somebody elects to do on a private money basis. So we just need to keep that in mind, too, in where we put these different kinds of statements in the body of the document.

MS. BERRY: Martin, did you have a hand raised?

MR. DANNENFELSER: Yes, I think there is a question. I guess I had a question where it says "clinical" there at the bottom. Is that synonymous with therapeutic? I guess based on the dialogue that Reed had there, I gather that that is what is meant there,

clinical versus informational benefit.

I guess just in general, I think this is a real touchy area when you get into the area of whether it is therapeutic. Is it therapeutic to the infant and so on. I don't know that you are necessarily going to have a situation where the government is going to recommend something to private insurers that it is not going to pay for itself. That would seem to be not in synch for the government to do that.

MS. ZELLMER: I also would just point out, there may not be any immediate utility to carrier testing, but I don't think that is something that we should discourage coverage of carrier testing.

MS. BERRY: Barbara?

MS. HARRISON: I think likewise, if we go with the Huntington's example, you can't do anything about Huntington's now. I think you'd argue it doesn't have much therapeutic utility in the sense of before you acquire symptoms, there is not too much that can be done.

Yet I wouldn't necessarily ? - that's just informational for personal use. I think that is the whole purpose of a group like this who could make those distinctions, you know, of Huntington's versus the sex of the child, which, kind of just depending on what your arguments are, could put you one way or the other on them. I think that is the purpose of a group like this, and not for us to be trying to flesh out what is appropriate and what is not.

MS. BERRY: Brad?

MR. MARGUS: It's only going to get more complicated. The other thing is we're talking kind of with the assumption that a genetic test is going to be an on/off answer. If we are talking about multi-genetic traits where the answer is going to be that you have a 64 percent risk instead of a 30 percent risk, now you're saying it is going to get even more complicated where people are going to wonder what the utility is of knowing it. This whole area is going to get messier and messier, as far as if you are holding it to a standard of it is definitely actionable or not.

MS. BERRY: Agnes?

MS. MASNY: It is not only going to get messier, it already is. I know in the oncology arena, we have already been faced with calls from people where genetic tests aren't being made available to the public. One was the case for ovarian cancer where they were looking at a panel of proteomic markers, and luckily the FDA stepped in, and at least the criteria there was that there was not enough validity studies that were done yet that could conclusively say that yes, this test is ready to be provided as a screening tool.

So I think that some of these general principles that we're trying to come up with, even if we have things like that, how many validation studies are necessary to show the effectiveness of the tests that could then move it into the public realm.

Secondly, just recently as a test for looking at modifier genes that will be used for the general public. Not just for high risk, but for the general public to determine what members of the general public will be at risk for breast cancer. So they are already out there.

MS. BERRY: Reed?

DR. TUCKSON: Brad's comment is actually terrific, as well as Agnes'. I'm just starting to wonder. Obviously we cannot solve this. I'm just sort of thinking in my mind, what needs to be different a year from today so that we're not sitting at the same table going, oh my gosh, somebody ought to do something about making this make sense.

I guess it would be, at least in my mind, I would love to frame the issue even tighter that says here are the set of conundrums that are before the country in this area. This is why this is new, and this is what the paragraph I think tries to do. It says that there is something new about these genetic tests that introduce a new level of uncertainty and complexity that is different than before.

As a result of that, there are the following kinds of decisions that have to be made. They involve these categories of issues. The current organization of knowledge that solves those problems is comprised of the following groups. They are inadequate. As a result, there needs to be something else immediately put in place that permits this work to go forward in an organized way.

I guess that is what I'm not sure about. I think what we ought to be calling for here is the body of people, the right agency that it needs to be located in to make sure that we're not here a year from now having gone nowhere. I think that is what this recommendation ultimately is trying to say.

Maybe not being so prescriptive about what the group does, as much as ? - well, we need to be prescriptive. I guess I can't take it any further than that. Is there anybody in government today that is charged with thinking these issues through for the government? I mean, who is in charge of this? So when Brad says it is 60, what happens when it tests at 64 percent versus 20? I mean, who thinks about this?

DR. KHOURY: Part of the problem here, Reed, is that the efforts are fragmented throughout our sister agencies here. I mean, we all have a piece of the elephant. That whole elephant needs to be constructed in a way that the whole is bigger than the sum of the parts.

I think, you know, coming back to, was it you or Ed that mentioned about this HHS-level czar or czarina to try to put it together. You know, we have fragmentation.

That's enough from my side, I guess.

MS. BERRY: And Reed, do you feel that it should be one agency or governmental entity? Or does the group think that perhaps we might consider some sort of task force or commission that has the relevant agencies involved, but also the experts from the private insurance world and other stakeholders?

DR. TUCKSON: Boy, it's a great question. I'm really influenced by what Muin has said. That is that it sounds like this needs to be at a minimum across HHS. I'm not sure, I'd have to think some more about whether or not I ought to invite private sector people to it or not.

MS. BERRY: Ed?

DR. McCABE: I was really quoting our esteemed leader when I brought that up before, who had made the comment in the past. I think Hunt Willard might be in a position to comment on this, because he has taken a position of leadership in a university where genetics and genomics used to be a department, and now has been elevated to a higher leadership role at Duke University.

I don't know if you could comment on whether that is for purposes of integration across the university. I think that's the kind of thing. We're looking for integration of genetics across HHS, and perhaps Duke is a model.

DR. WILLARD: Well, I rarely conclude that academia is a good model for government to follow. The argument that there needs to be an integrating body, whether it is a

person or a body, is a different question. I think that is a recommendation this group may take very seriously. This isn't, as our mantra at Duke is, this is not just science anymore. This is science and policy together. So you can't point to any one of the HHS entities specifically and say, you're the one who should be in charge. They are the ones who should be in charge of some part of that pie, but then there is no one actually watching the entire pie.

So it is a reasonable recommendation for us to debate. Although I personally always hesitate to recommend yet another level of bureaucracy above the existing levels of bureaucracy. In this case, a coordinating body or a coordinating office, maybe there is something to be said for that.

MS. BERRY: Steven, did you have something?

DR. GUTMAN: Yes, I wanted to speak to Muin's comment, which is that it is a very colorful mosaic of regulatory controls in place that actually are driven by very different statutory bases and very different cultures.

You actually have as a baseline a very broad coverage by the CLIA program, which looks at the analytical validity and the underlying quality system in place. You have FDA for the products we review, which looks at analytical and clinical performance, at least in terms of a surrogate outcome on a device-by-device-specific basis. Then you get CMS, Aetna, or Blue Cross/Blue Shield, or Kaiser and whoever else, to actually pay for the damned thing.

In that case, they actually ? - I don't know that they will all uniformly follow Dr. Rollins' suggestion that before ordering a test, you decide what you'll do if it is positive, and what you'll do if it is negative. If you do the same thing, don't order the test. If you don't know, find out before you order the test. I don't know who does that or who doesn't do that.

I do think when the rubber hits the road, and when CMS makes payment decisions, and I presume the same is true for Blue Cross/Blue Shield, Aetna, Kaiser, and others, that there is the introduction of some kind of cost-effectiveness determination, or some kind of utility determination, even if you don't actually know the utility, which perhaps transcends both CLIA and FDA.

MS. BERRY: Alan, and then Reed.

DR. GUTTMACHER: Yes, I think this is in some way sort of a multivariate analysis, and therefore, very difficult. It also brings to me the question of sort of genetic exceptionalism in a strange new application, perhaps. That is obviously I think everyone who would be against coordination, I mean, are the same people who are against motherhood and apple pie, I suppose. But besides that, coordination makes a lot of sense.

The question is if you tightly coordinate, first of all, the questions would be sort of regulations and laws that would allow you to do that are questionable. But also if we go back to the infectious disease analogy, we certainly don't have a czar or czarina of infectious disease within the federal government.

Now, maybe you would argue that we'd be better off for that, looking at recent problems with immunization, but I'm not sure that we would be in some ways having a multitude of different folks coming at this from different perspectives, even within the federal government, is a good idea.

Coordination obviously would be a good idea. We want to favor that. But the question is how do we favor coordination within what is sort of allowed. There is some

difficulty in creating completely different ways of dealing with things which "are genetic or genomic" versus everything else, whether it be in science, medicine, or whatever.

Then there is also I think the question of are you going to coordinate things without making monolithic kinds of things. So it is a difficult kind of juggling act.

MS. BERRY: Reed, and then Muin.

DR. TUCKSON: Well, we have a lot on the table here. First, I think one good model for how everybody benefits from what government does, and public resources and services to the nation, is the U.S. Preventive Services Task Force.

Now, here is a place where, again, I know everybody understands what that is. But to make sure that we have the best scientists that we can find in prevention, you know, who look at the literature carefully and thoughtfully, analyze that in a publicly transparent way, and make very specific recommendations.

Then those recommendations are available for people outside of government to benefit from and make decisions based upon it. It is a terrific public service, and it is a good use of tax dollars, in my opinion.

So if you take that idea and you say here is a special new area of concern, I would sort of be saying, I don't think, Alan, that we need to necessarily create another bureaucracy. Maybe it is that we ask the government to bring its best thinkers together for a task. The task is we identify that there is a challenge that needs organized thinking across CLIA, AHRQ, and CDC. The task is that there are some specific questions around this new technology, these new interventions, that are different, and therefore require some thoughtfulness.

We want you to use the federal resources to bring it together transparently, and then make that information available for CMS and others to be able to take advantage of. We define it very specifically in terms of the range of issues we want, let the government figure out how to pull those people together. Don't create another bureaucracy.

When they finish their task, they all go home, and if they need to revisit it at some period, refresh it periodically, that's for them to decide. But then everybody has the benefit of it. That is one idea for you to shoot at as a way to go.

MS. BERRY: Muin?

DR. KHOURY: Yes, I think this is kind of the model that we've adopted with the EGAPP initiative. We've had a lot of discussion with AHRQ before we launched the EGAPP initiative. The U.S. Preventive Services Task Force is the gold standard.

The problem, as Linda Bradley presented yesterday, is that the U.S. Preventive Services Task Force focuses a lot on clinical utility and the primary care setting. If you were to do an analysis of most genetic tests using the strict criteria of the U.S. Preventive Services Task Force, most of them will not meet that threshold.

As you said earlier, Reed, this is a complex new arena. We hear about that information for the sake of information, i.e., clinical validity, like you have a diagnostic test. It could have clinical utility built in by knowledge of your diagnosis, because it can avoid diagnostic odysseys. But then you bring in the ethic and legal issues, and then you have an ACCE elephant sitting in the room.

DR. TUCKSON: Muin, let me just make sure. I don't want to monopolize this. I want just for clarity sake, you are absolutely right. I do not raise the U.S. Preventive Services Task Force for any other reason other than their social role.

DR. KHOURY: Right.

DR. TUCKSON: Of organizing best thinking through use of public resources and making available to inform what government does in those outside of government in the interest of the nation. By definition almost, what this task is is the antithesis in terms of the methodology.

The U.S. Preventive Services Task Force, if it doesn't have 18 bazillion articles, they don't rule. So this is almost the antithesis in the sense that this would be going to unchartered waters, whereas the U.S. Preventive Services Task Force only goes in well-navigated waters.

DR. KHOURY: Can I take on another issue? The issue of genetic exceptionalism. I guess also Linda presented on this yesterday. I would agree with you, Alan, that there is no need for an infectious disease czar in the 21st Century.

But imagine infectious disease at the beginning of the 20th Century, or the 19th Century, where the technology was still new. This is what we're facing with genetics. I mean, I don't know historically whether any one of the HHS was charged with controlling infectious diseases in the country, but I think the CDC comes as close to one agency that was tasked with ? - actually, its name was Communicable Disease Center back in the '40s or '50s when it was created.

So I think the model really applies only to the extent that you just have to subtract 100 years. If you look at genetics 100 years from now, then there is no need for that kind of coordination.

On the other hand, I agree with you. I don't think we should treat genetic tests in such an exceptional way per se, but the issues of the magnitude and the complexity of these tests deserve a look. Otherwise, we won't have all these advisory committees that have been formed, from SACGT up to SACGHS. There is a special one on newborn screening.

The government and the private sector have decided that genetics is worthy of a special look. I think the principles that we're talking about as we move forward in the practice of 21st Century medicine or genomic medicine, that will be integrated and can really have a long way in terms of influence on the practice of medicine in general, whether or not it is genomic or not.

I agree with you. In the long run, we want to have genomics as an integral part of the practice of medicine. But how to get there is sort of the challenge that we have right now.

MS. BERRY: Emily?

DR. WINN-DEEN: I sort of want to agree with Muin, but go a step further. I think the issue is really what do you do with emerging new markers? I think it is broader than just genetics, except that genetics is probably the vast majority of emerging new markers.

Anytime there is a new candidate marker for something, you go through this process of gathering evidence that this marker actually has some usefulness. At some point in time, we need a group that gets together and says, okay, we agree that this is ready, and it should be adopted. I think that is part of the thing that is behind this recommendation is how do we get there, and the fact that a lot of these new markers are going to be in genetics just gives us sort of an opportunity to put a group with some special expertise in genetics together to do this.

But I think we're sort of talking around this and not really getting to the

point of making a recommendation. I personally think the recommendation is not too bad, except that I would take and change the word "could" to "should." "The Secretary should task an appropriate group to develop a set of principles for coverage decisionmaking for genetic tests." That would apply to both employers doing private insurance and to public health insurance.

I also think that that group needs to have representation, and not just from people in the government. If it is going to develop recommendations that would apply to all, that you have to build consensus that everyone would agree that when this group makes a recommendation, that everybody is going to buy into it. You can't get that unless you have the stakeholders from the private sector involved as well.

MS. BERRY: Emily, are you talking about, and are we as a group talking about the group specifying when a specific technology or service should be covered? Or would it take a step back and be a little bit more vague or broad and establish principles and say these are the principles that we think everybody should apply, and then in going through that process, they'll make their own individual determinations as to whether they will cover something or not?

DR. WINN-DEEN: Well, I think to establish a set of principles is a good and useful task for everyone. I think SACGT tried to do that, and they went through a whole big algorithm of when do you know that something has reached the clinical utility threshold.

There is still a lot of gray zone there. A lot of medical specialty groups have sort of stepped up and said, we're going to make a recommendation within our disease area specialty that this test is ready, and it should be applied in the following ways.

So it is a very fragmented thing right now. The question is just should we as a group make a recommendation that the Secretary of HHS somehow centralize this function and create a group that at minimum creates a set of principles, which then could be used, and potentially if you read the whole recommendation here, it goes farther, and actually says okay, and of the tests we know today, here are the ones that are definitely not on the list, here are the ones that are definitely on the list, and here are the things that are, well, basically everything else is still in the gray zone.

It either hasn't been evaluated yet, or there is not enough body of evidence, or whatever, to put it in one bin or the other. But that's the only way you're going to get to being able to make good coverage decisions and have some kind of unified coverage of new tests.

At some point they cross the threshold where just some people are covered into we really believe this is medically useful, and all carriers, public and private, should be paying for it.

MS. BERRY: To kind of tie this up, I guess is it safe to say that the group feels that there is something special about genetics and genomics that sort of cries out for some entity trying to provide guidance to either the public sector or the private sector and others? I mean, that is sort of the first question. Do we feel that there really is a need for this guidance?

Ed?

DR. McCABE: Yes, I would argue there is a need for guidance. I would argue that there is not a special need for the guidance, but we aren't tasked with developing guidance for infectious diseases. We are tasked with developing guidance for genetics. So that is what we do. But I would argue it is not special, but there is a need.

MS. BERRY: Then what would be the most appropriate body to address that need? Would it be HHS and leave it vague? Would it be convening a task force that includes public and private sector stakeholders? Would it be a particular agency? Do we want to recommend the body, or do we keep it as is where it says HHS will convene this group, and we don't specify any other details?

Ed?

DR. McCABE: In the original, it says that "HHS will task a group," and I actually prefer that language than "establish a group." In fact, the groups may already be established.

I would also maybe even make it vague and task group(s) with the "s" in parentheses, because there may be a need for more than one kind of a group, as we've heard from ACCE and EGAPP. It may not be a one size fits all.

MS. BERRY: Reed?

DR. TUCKSON: I would agree with Ed, in that if I understand his point, first is that it is tasked instead of established. Therefore, it doesn't look like you are creating a whole series of reexisting bureaucracy that has a life of its own, which I think is something that we ought to be completely transparent about.

Number two, we want to be very specific about what the problem is that we want to get solved, but leave it to HHS to figure out how to best do it. Because as we've heard from Alan, Steven, and Muin, we can't possibly try to figure out all the machinations of the doggone government. That is only the things that they can figure out how to best use their resources, and who ought to be the charge of it and so forth. If those are the points that Ed is making, I endorse those.

MS. BERRY: Then to take it to the next layer, we think there should be guidance. We need guidance. There should be a group tasked with producing that guidance. Leave it to HHS to come up with the appropriate group or groups to do that work. Then the next layer is what is that work?

If it is establishing a set of principles and general guidance, that is a group then that could do the job and then fold their tents and go home. If it is to do that plus analyze specific technologies and be in existence and perpetuity as new technologies and services come into existence, there is some entity there that passes judgment on them, that's a different story. That sort of points us in the direction of an entity that has to continue to exist.

DR. TUCKSON: What if we were to give them the opportunity as part of their charge to determine what, if anything, needs to be done after they have, you know, done their work. Maybe make them make the recommendation back, instead of us trying to predict whether or not ? - because I think at this point we don't have enough information. To put it to their charge to make them determine what is the most appropriate course of action, and make that recommendation back to the Secretary, and to us.

MS. BERRY: Debra?

DR. LEONARD: Well, it is one thing to develop the general principles that could then be applied to tests X, Y, and Z, or medical conditions X, Y, and Z. We have a whole bunch of recommendations to provide the evidence base to Medicaid, Medicare, and I don't think those are going to be communicated as general principles. So it has to be done on a test-by-test, disease-by-disease basis, so that the programs can actually change what they're doing, what they're covering, and what they're reimbursing.

So I don't know whether this group would do that, but I would see a series of recommendations. One where you establish a group to do the principles, but then those principles have to be applied to create the evidence base, and then that evidence base is what is distributed to all the people who need to know that information.

DR. KHOURY: One of the things we learned in the ACCE project is that you are dealing with a lot of apples and oranges, and we have only done five systematic reviews from prenatal testing, carrier testing for CF, all the way to BRCA1 and hemochromatosis.

At the end of the day, I have to agree that a general set of principles will not be sufficient. As a matter of fact, it won't be difficult to come up with this set of principles given all the work that the previous committees have done. The application of these general principles to a test-by-test basis is going to be complicated.

It is not only a test-by-test basis, but by intended use. You can use the same test for either carrier testing, symptomatic, or presymptomatic. Then all the parameters will change in terms of validity and utility, and then some of the ethical issues.

So I think if we have the U.S. Preventive Services Task Force as a gold standard for how those things are done with evidence-based principles in mind, then we shouldn't short, I mean, we shouldn't sell this very short. I think each one of these test evaluations is going to involve a synthesis and integration of the available literature, both published and unpublished.

These things are not cheap. I mean, it may take sometimes six months to a year to evaluate systematically what is going on with a specific test. I mean, for me, it is not rocket science to figure out that whatever you task HHS to do will have to go beyond coming up with a set of principles, but developing the approach and methodology for how these principles can be applied to specific situations.

MS. BERRY: Well, I've gone ahead and fast forwarded to the evidence-based recommendation. We'll go back to the others. I wanted to pose the question to the group, should this be woven into the earlier recommendations so that we task some sort of entity, HHS tasks an entity to come up with a set of principles. That provides the general guidance. But then as Muin just mentioned, and Debra, there may be a need to go further on a technology or test-by-test basis.

That is what this recommendation gets to, which is assessing the actual evidence for specific test and technology. This group would also be charged with that task as well. Do we want to have a two-part component to the charge for this group?

DR. LEONARD: I would argue that there is no one group that could do all the tests. So I think for each test, you may have to have a different group with different expertise, I don't know. Once you have the principles, I think there is another gap, which is how do you determine which tests to do this for?

Are they the one with the greatest public health impact? Is it the one with the greatest penetrance in the population? Is it the one with therapies? I mean, how do you generate ? - that would be a very complex process, because you're going to do these one at a time.

It will take time to do that.

DR. KHOURY: You know, I wish AHRQ was here, so I'm speaking on their behalf. Forgive me on the webcast. One of the principles of the U.S. Preventive Services

Task Force is that you have this independent body of 10 to 15 people that meets three or four times a year. They can decide, people can come to them and say okay, let's review this evidence for whatever it is. Whether aspirin prevents heart attacks, or whatever the issue is.

Then they deliberate. They are independent, and then the Commission in an evidence-based center, and there are many of these centers around the country. Obviously people who are specialists in cystic fibrosis may not be the same as in BRCA1. Then they do the evidence-based analysis and they bring it back to the table.

The task force, what they do is they look at the evidence, and they make the pronouncements up or down, quality and quantity of the evidence. I guess the experiment we're doing with EGAPP is sort of a collaboration with AHRQ to define the methodology, and then simulate that principle of an intendant body that these were the stakeholders, and then commission the systematic reviews, bring them back to the table, and then make some kind of pronouncement.

Then more importantly, because I think we feel that this is an area where a lot of research will have to be done, is identify very specifically the gaps in our knowledge then that can be funded through both the private and the public sector. So that a year down the road, there will be a change in the recommendation, or at least a statement about what we know about the genetic tests.

We won't get stuck for years and years saying that this test is no good. But next year maybe we'll come back and reevaluate it based on new data. So I think this is a moving target in a lot of ways.

DR. LEONARD: Muin, how do you see what we're proposing here as different from EGAPP, and should we just support EGAPP and ask the Secretary to give that resources?

DR. KHOURY: No, I don't think you should endorse specific activity. You should ask the Secretary to get the agencies to get their act together and see who is doing what, and let us collaborate. Because there may be another prescription to do this. I cannot presume that CDC has the only valid way of doing this. I think it is better for you to stay over maybe 10,000 feet, rather than go down on the ground with us.

But by convening, by asking for it, then the Secretary can poll the agencies and get them together and say okay, who is doing what, and let's figure out how to do it best.

DR. TUCKSON: As I'm listening to Debra and Muin, I am getting more and more convinced and confident that, you know, we have assessed that there is a problem. Here is the problem. We have assessed that certain things are going on, but they are not adequate. Therefore, this needs to occur. You need to make this happen. We want this done by a certain time period. It needs to be done.

I think that is really what it keeps coming down to.

But James, I wonder on Number 1, let's just say for you guys. Don't you do this?

DR. ROLLINS: Yes.

DR. TUCKSON: And I guess the question ultimately becomes, that is what you do? It is what is lacking? Or what don't you have that would make your job be more effective in this new area than you have today?

DR. ROLLINS: Yes. Everything that is mentioned in Number 1 is currently what we do when we do assess a new technology, be it some procedure, be it a new

surgical technique, or even a laboratory test. So those are the processes by which we establish something that has shown sufficient evidence or has not shown sufficient evidence.

DR. TUCKSON: So the question it comes back to is can we articulate, and I'm back to where Debra was on this, I think. Can we articulate the set of issues that are of concern? The set of things that require new principles. The set of conundrums that are unique to this genetic era. Can we define with greater specificity the confusions brought by predictive tests that may not have clear clinical correlates?

Can we define with precision the cost-effectiveness decisions around introducing new technology when in fact there may not have been any preexisting method of intervention before with which to compare whether this is more cost-effective, too. If that is even English.

So I guess what I'm wondering is if we can sort of start to figure out what would be that list of concerns, and then frame those back and say here is what we're talking about as the range of issues that we think that this group needs to look at, and that you, Jim, and your agency, would benefit from having that information pulled together so that you can in fact through your existing processes make the decisions that you need to make, and such that information can be made available to the public transparently for others to take advantage of it.

DR. ROLLINS: I think ? - I'm sorry.

MS. BERRY: Pardon me. Go ahead.

DR. ROLLINS: I was getting ready to say, some of the things that you requested, such as cost-effectiveness and things on that order, that is something that we have actually not been charged to do. But I'm sure that information can be derived from some other source which could be used to supplement decisions.

But in terms of some of the other things which you've recommended, I do think that based on what it is that we currently provide, that additional information can supplement us in terms of providing additional information, or providing additional information for other uses.

MS. BERRY: We have to keep in mind as well though that we are also talking about private plans. Some plans have very, as identified in the report, very elaborate guidelines and processes that they go through, and have been willing to share that publicly when they make these determinations. Others may not be so transparent.

So to the extent it may be that CMS has this process in place, but maybe in the private sector it doesn't exist, or some do and some don't. So there may be some value in having someone outline that so that the general public will be aware of what goes into a coverage decisionmaking decision, and it is not just some hole that, you know, the request is in and no one really knows how the decision is actually reached.

Hunt?

DR. WILLARD: I'm afraid we have to do a somewhat better job of articulating what the problem is. I think we probably all somewhere in our guts think that there is a problem.

If I just heard from Dr. Rollins that CMS is doing Number 1, then I either need to hear that CMS isn't happy with the job that they're doing, or that someone else isn't happy with the job they're doing in order for me to feel comfortable to say okay, now we've identified a gap. We either need more information provided to help CMS, or more information to have them come up with a different answer than they might have come up with.

That same question could be asked over and over again, whether it is for the CDC or any of the other groups that are within the federal government, or under HHS. My concern is we just are not drilling down to a very concise statement of what the issues are, other than there is uniform angst. I think we probably need more than that.

MS. BERRY: Dr. Rollins, I know that CMS undertakes this type of process in Number 1 for all medical services and technologies. Would you say that your ability to undertake that process in the area of genetics is hampered in any way because of a lack of evidence? Or do you feel that CMS has what it needs to proceed with that process in this area?

DR. ROLLINS: I think that CMS is capable of pursuing any request pertaining to the effectiveness of genetic tests. So I do think that as long as there is information out there available to assess, we are in a position to make those decisions.

MS. BERRY: Well, then Hunt goes back to the original threshold question which we raised, which was is there a uniform feeling that guidance is needed, that there is some issue, problem, or deficiency, in the fields of genetics and genomics that cries out for some sort of guidance by a federal body or federally tasked or charged body?

DR. ROLLINS: I'll make a quick comment. When looking at a technology, not only with CMS, but also the commercial plans in terms of coverage decisions, they base their decision on the effectiveness of that particular technology.

If there are other questions which go beyond that, such as cost-effectiveness, or certain types of utility, that is something that is beyond what it is that we look at. We essentially look at the literature in terms of determining whether or not a test is effective or not effective.

So any additional questions beyond that might be something else that might, you know, we may not address, or other plans may not address. For that reason, additional information may be needed, or additional direction might be needed to address those additional questions.

MS. BERRY: Agnes?

MS. MASNY: I just have a question for you, Dr. Rollins. If a test though was deemed to be, a screening test then would not be covered by Medicare or Medicaid. Would then CMS undertake this type of assessment?

DR. ROLLINS: We do cover diagnostic services, as we discussed yesterday, in terms of screening diagnostic services. That is something that has not been required, it is something that has not been mandated by the government for us to do. As I say, diagnostic tests, yes.

MS. BERRY: Hunt?

DR. WILLARD: You know, in essence, the issue comes down, and I'm not sure whether we can do it, that either we have to, or we need to charge or recommend to the Secretary of HHS that he attempt to do what the Supreme Court refused to do for pornography decades ago, which is to try to define exactly where the gray zone is, which is almost oxymoronic. By definition, you can't do it.

I mean, we all agreed going around the table that testing for Huntington's disease in certain settings had value. It was effective at some level. But testing for blue eyes or brown eyes was not going to be of value, or not viewed to be effective, and therefore wouldn't be.

So those are polar, but the area in the middle, unless we feel we can bring

something new to the table, or that some other group is going to bring something new to the table to define better the gray zone, I'm not comfortable that we have much to add to the dialogue, except acknowledging that there is a gray zone, and it is going to take great care and thoughtfulness on many people's part to continue to evaluate this over time.

MS. BERRY: Remember, we were talking about two different parts to the charge. The first was a set of principles which would look at issues such as the informational utility, medical effectiveness, the preventive nature of genetic services, and have these general principles developed so that they could serve as a guide both in the public and the private sectors.

The second part was more the test-by-test, technology-by-technology assessment based on the evidence. It sounds like that second part may not be a problem. CMS feels that it does that, and can do that. There is a question about whether the private sector uniformly does that, or whether there is some need for guidance there.

But if we're going back to the beginning, Hunt, to you suggesting that even the first part of the charge that we talked about where we would have some group establish a set of principles, that maybe that isn't clearly necessary.

DR. WILLARD: I'm certainly open to the possibility that a group could come up with a set of principles that would say, this is what we mean by the 5 prime N, and these ones are clearly at the 3 prime N, and then there is all this stuff in the middle that we can't really declare.

I mean, if we felt that there was a group that could provide sufficiently robust and specific guidance on what is at the left and what is at the right, then there would be some value to that. But if it is simply to discuss year after year, as Reed was alluding to previously, everyone saying boy, there are some things on the left, there are some things on the right, and there is a whole lot in the middle, then I'm not sure if it is worth either the recommendation or the dollars that would flow therefrom to pull that group together.

MS. BERRY: Brad?

DR. TUCKSON: Let me give a process check, Cindy, as you do this. We're at 12:07. We sort of end this part at about 12:30, allowing us to do a working lunch and break in there and all that, and start at 1:00 with the testimony from the public.

So Cindy, you've got a few more pages to go through as well, so I think we need to try to figure out how to nail this one down. So I think all the comments from this point on need to be what is the solution to how you all want to get out of this.

DR. LEONARD: Reed, are you working from an old schedule? That's not the schedule that is in our folders.

DR. TUCKSON: Let me see. We've got this idea of sum up and decisions and plans for gathering public comment around 12:30 to 1:00. I'm basically told it is sort of like a placeholder for finishing this. That is why I'm saying we have until basically around 12:30. Then we have this working lunch which we've got to do. Did I miss something?

PARTICIPANT: We're going to take a break.

DR. TUCKSON: Well, of course. Why would you call me to task? I absolutely have scheduled a break. We don't need to take a break and then go to lunch.

We're going to go to 12:30, because we've got to get this done. Then we can go take the break and get the lunch, and then come back and do public comments. Okay?

PARTICIPANT: (Inaudible.)

DR. TUCKSON: No, we're going to take the 10-minute break, and then we're going to bring the lunches back in and we're going to keep talking. You've got to talk while you eat, or otherwise you've got to pay money for your lunch. We've got until 12:30 to bring this to closure.

MS. BERRY: Brad, then Emily.

MR. MARGUS: My suggestion was maybe if we're concerned about tests that might not be covered, and particularly with CMS, just to clarify what Dr. Rollins said. If there is information available, then CMS reviews it and uses it.

So maybe we could in situations where information is available, that CMS would speak up about it, or that we would be that someone then fills in. What we're concerned about is when, and correct me if I'm wrong, is that we're concerned that in certain situations tests fall in La La Land and don't get covered because there hasn't been anyone building a case for it, and CMS is very open to reviewing information and using it if information for, you know, necessity or validity is there.

That's only when the information is there. If the information isn't there, what do you do? Maybe we should put that the recommendation would be if there are gaps to having that information, then something has to happen.

MS. BERRY: Well, and also, to go back to a point that was raised earlier, CMS wouldn't undertake the whole process like this to look at the evidence for a technology that it is statutorily prohibited from covering. So we can't completely rely on CMS' ability to be the final arbiter here.

They'll do what they can within the purview, within the scope of their authority, but then there are other payers and components of the health care system that do require some sort of process like this that can't just be CMS because they are not bound by the same statutory constraints.

DR. ROLLINS: I think that most payers, commercial as well as CMS, follow this same type of process. So in terms of evaluating the evidence, we are all pretty consistent. The only restriction is, as I say, because we do not cover screening tests, those are something that we would not review. Commercial insurers, they have the option of covering both diagnostic as well as screening tests. As I say, whether or not it is diagnostic or screening, they follow the same type of evidence-based review process.

MS. BERRY: Emily, then Debra.

DR. WINN-DEEN: Okay, so in the interest of moving this all along, pages 60 and 61, and then this whole box that we're discussing on page 69 are repetitive, and we should get that organized in such a way that we don't say the same things twice, and don't have the recommendations on two separate pages.

It seems to me that what is lacking is not that there is a process that happens at CMS, or that there is a process that occurs at different private insurers, but what is lacking is sort of a publication of what that process is. So that people working in the field with emerging tests, many of which are these genetic tests that we're talking about, know exactly the questions that are going to be asked, so that data can be generated to address those questions.

So I would say that maybe one of the things we could ask the Secretary to do is to ask CMS to publish in some manner, if you don't already, that list of questions. What are the criteria that you use that you go through in the process? If you have a standardized process, what is it? Can it be transparent?

And then the second part, which is this whole issue of RFAs, is I think a way to address the issue of lack of evidence. A lot of things are in the gray zone because we just don't have enough information yet to say that they belong in a black or white category.

So I personally would like to advocate for continued NIH kind of RFA grant support to continue to generate information. Maybe it is CDC studies or whatever studies are required to get things to the point. We have enough evidence to say this is right, this isn't right.

I think that the hereditary hemochromatosis case study that is on the opposite page there is maybe a good example of that, where clearly you can test these mutations that they have some utility in clarifying a diagnosis in someone who is presenting with family history or signs and symptoms.

We don't yet have the body of evidence to say it is worthwhile doing a screening, population screening, and so we are doing a big study funded by the NIH to answer that question. So I just would like to sort of bring this to closure. I think the gap is that we need clarity on what the criteria are that generate coverage and reimbursement decisions, and then we need a way to fill the gaps for new things so that you can generate the evidence that's required to go through that process.

MS. BERRY: Suzanne?

MS. GOODWIN: Actually, the Medicare Prescription Drug Act tasked the Secretary with making available to the public the factors that are considered when making national coverage decisions. I believe CMS has taken the first steps in that process. So it is not available yet, but it is something that is being done at the moment.

MS. BERRY: Debra?

DR. LEONARD: In addition to the preventive aspect that is not covered by CMS, I would also argue that what would be covered by Medicare with an over 65 population is very different, and may not be considered for the 5-year-old, the 20-year-old, and the 40-year-old. So I don't know that CMS' policy coverage decisions apply to the entire population that is going to benefit from genetic testing as opposed to being restricted to the elderly population.

DR. WINN-DEEN: I guess I just meant that there must be a list of questions that they go through, and that set of questions that they go through, aside from the fact that one of the questions may be is this a screening test, yes or no, and if the answer is yes, it is off the table. I think that the list of questions in the process would be informative for most tests, probably.

DR. LEONARD: Except how much do you take into consideration with these decisions that you're talking about the over 65 population or the elderly population, as opposed to what would be good for a 20-year-old?

DR. ROLLINS: Well, I would respond and say that about 85 percent of the Medicare population are persons 65 and over, and like 14 percent for disabled persons. So it would apply to that group.

In terms of a process, CMS does currently have a process, and a person can actually go to the CMS website basically telling them how to initiate a process in terms of requesting a national coverage decision for a particular technology.

That information out there also would help in helping them to determine what type of information is necessary for that process to take place.

MS. BERRY: Barbara?

MS. HARRISON: I don't want to get us into another whole topic, but I was wondering, we have been talking a lot about testing, but we haven't really been talking about the accompanying services to testing, i.e., genetic counseling. How decisions are made about whether that is covered, or not, or even in whether a physician feels that he could bill for that or not, since we know genetic counselors can't.

I'm just wondering, would it be a similar process if someone was trying to pursue coverage for counseling?

DR. ROLLINS: I don't know, but I would think it would probably be a similar process.

MS. HARRISON: Okay. Well, just while I had the mike, the other list that we're working with where we read out some of the principles that that group should work with, I wanted to propose to stay with that, which I'm not sure if we're going to.

If we do stay with it, I thought maybe something to the effect that the principles should also address accompanying services necessary like genetic counseling or genetic evaluation to ensure quality care and recommendation of tests. Just to make sure that there are tests that can be available, but some tests should probably only be available if they are recommended by a genetics professional as opposed to a general physician, nurse, or something else.

MS. BERRY: Reed?

DR. TUCKSON: Yes, actually in danger of violating my all comments must lead to conclusion statement, Emily, I'm glad you brought us back to this hemochromatosis case study. Can I just ask, does anyone remember where the groups, the working groups, were they in government or out of government that created these conclusions? Does anybody know where that came from?

MS. MASNY: I know that there was a paper from the task force on hemochromatosis. So I think one of the aspects of this is from the United States Preventive Services Task Force.

DR. TUCKSON: This was a U.S. Preventive Services Task Force?

MS. MASNY: Yes.

DR. TUCKSON: I mean, if you look at this, Emily, I think it is really right to bring this out. Here is a great example. Here is a complicated issue that has a significant prevalence and penetration in society. There are a lot of people involved, you've got a test for it. The question is, do you offer this or not?

Then they make a very cogent sense of a clear evaluative process that ultimately concludes that under which conditions this is appropriate, and which conditions it is not appropriate. Having this kind of analysis available for decisionmaking is extremely important.

How do you organize yourselves to be able to create this kind of analysis on a test-by-test basis? But if this came out of government, I'm just wondering what the organization of government services are that made this happen.

MS. GOODWIN: Part of this case study was included based on a conversation at our previous meeting where I think you brought up this example. Are you familiar with the working groups?

PARTICIPANT: I'm sure I was at one point.

DR. WINN-DEEN: I have all the papers in my office. I can send you them.

DR. TUCKSON: No, I'm sorry. I just was trying to see whether or not this was a model of a way of proceeding.

James, is this the kind of information that if you all had available, you could make decisions?

DR. ROLLINS: I think that type of information would help supplement us in making a decision, yes.

DR. TUCKSON: Yes. The question is if you look at the genetic tests that are out there for you to have to make decisions about today, do you have a place to go to get this kind of data?

DR. ROLLINS: We review the peer review literature. We would also review any information available on some of our public websites, such as AHRQ. We sometimes communicate with various societies to get a perspective on the utility of the test.

MS. BERRY: Muin?

DR. KHOURY: I guess I was out. Was a question specifically posed to me? Or shall I just jump in?

DR. WINN-DEEN: We just wanted to know what the background was on HH, if you remembered if that was all government, or if it was public/private.

DR. KHOURY: No, I mean with hemochromatosis, we've had a number of meetings, starting with one we held collaboratively with NHGRI back in 1997 to look at evidence for population screening.

The most recent activity is the ACCE report, which is a systematic review of the whole elements of population screening. It looked at analytic validity, all the way to the ethical issues. That was put together by the Foundation for Blood Research in collaboration with people who are sort of the experts in hemochromatosis.

DR. TUCKSON: Muin, what I was trying to get to, and you may have answered it, is just simply a matter of this is in there as a case study. I was wondering, is this a case study of the way in which the system can work to organize data, information, and analysis, and then feed it back for decisionmaking for government and others so that you expedite the concerns that we had here? Is this a model that can work? Or does it really tell us that, I mean, the ranges of permutations and complications on any one of these. You can't create a freestanding body that is going to do it all the time, and that you basically just have to do it case by case the best you can.

DR. KHOURY: Right. I think this was brought to SACGT a couple of years ago as a case study for how government agencies work together. I think it is a good case study. Once the gene was found in 1996, two agencies came together and said okay, let's look at it. There were early calls for population screening.

However, having said that, I don't think, unless there is some kind of a situation where these things are anticipated, because there will be many, many hemochromatosis to come, pharmacogenomic tests, who is going to be keeping tabs on this? I think that is the issue. Is it a case-by-case basis, or something a bit more overarching?

MS. BERRY: Sarah?

MS. CARR: Well, what I was wondering about was if these are done in sort of an ad hoc way, how are the decisions made about what next issue to look at? Is that where some help is needed, that there be a body that would say okay, well now the next thing that we need to look at and evaluate through this sort of process is this test or this mutation?

Because otherwise, it is going to be decided I guess through specific people who are interested in that area. So I think that that is where the committee might consider some more systematic approach.

DR. KHOURY: Right now, there is no such process in place. It all depends on the sort of networking and the discussions that go on in the hallways and behind the scenes with the professional society.

Somebody might say, we have a test that we think is reasonable, but there is really no process that is established. We were hoping that the EGAPP would serve as a model for such a process. Maybe not the process itself, but over the next three years we can learn from it and see how it will work.

MS. BERRY: How about, just to try to wrap this up, to go back to the beginning. Is there a consensus that we do need some guidance in the area of genetics and genomics for public and private payers that would take the form of an entity or several groups tasked by HHS to establish a set of principles along these lines that would assist in making coverage determinations? That is sort of the threshold question. Is the problem of lack of guidance severe enough that it merits this group making a recommendation specifically to the Secretary setting up some sort of task force or entity?

DR. TUCKSON: I come back to the Huntington's point, Hunt's point. I'm just not sure that we have defined enough the problem. I just don't see how we can make the recommendation today until we all at least have a feeling that we can rehearse on the same gospel hymn, the problem. I just don't know whether we all have in our mind what we are trying to solve. Maybe I'm the only one.

DR. LEONARD: Could we ask the agency representatives to provide input if they feel there is a gap, what that gap is, so that it could inform our discussion when we come back to this next time?

DR. KHOURY: I'd refer you to Linda's presentation yesterday. Maybe I can go over it again. I think the case can be made again and again that there is a big gap right now. It is a knowledge gap, it is a policy gap, it is a reimbursement gap, and all of these things go together.

The number of tests that are coming down on the market are not going away, they are only increasing. With all due respect with the CMS work they're doing, I mean, there is a screening exclusion, and they are mostly dealing with old people. So we have to develop something a bit more sustainable, and I think that pulls all the agencies together.

DR. TUCKSON: Can you define, and I hate to put you on the spot.

DR. KHOURY: No, that's okay.

DR. TUCKSON: But can you define it? I mean, can you just say in your mind, you know, the preamble would be, Dear Secretary, we have discovered this problem. It is, therefore, do this.

DR. KHOURY: Well, you know ? -

PARTICIPANT: With the understanding that you won't be saying that. We will be.

(Laughter.)

DR. KHOURY: Well, let me step back here. If we are trying to develop a robust process for evaluating genetic applications and practice, and what we have to do, I mean, you as a committee have taken a look at issues from discrimination to reimbursement, coverage,

evidence-based, and criteria.

If you think that there is no problem, then we can all go home. At least what I said, I think that there is a major problem, because the number of genetic tests that are coming down the pike have no way to go but up. Each one of them, it defies our conventional wisdom of evaluating clinical utility in the traditional AHRQ or CMS model.

We are dealing with different parameters of information, ethical issues, and clinical validity, other than the medical way of looking at it. Thirdly, basically there is really no process right now in place that takes care of all these issues together. I mean, they are a fragmentation of efforts.

AHRQ has not up to this year, taken on any genetic tests for primary care. They tell me if we use them through the AHRQ U.S. Preventive Services Task Force model, they will all fail. Why they will all fail is because they have a clinical effect on us, and a traditional way of doing business that I'm sure the genetics community may want to modify to have a further look at this.

So there is coverage and reimbursement, and there is people calling for services. I mean, we heard from the consumers yesterday. So I can't reinvent the wheel. I thought that the wheel had already been invented.

DR. TUCKSON: So Muin, what it boils down to in terms of I hear you is we all understand the issue that there is lots of tests coming in. What you are saying is what is different is that we do not have a robust enough ability to evaluate the clinical utility of these new tests because there is something special about these new tests, i.e., they are predictive, and something else about them.

It has to do with, as you boil it down, the ability to evaluate clinical utility of a new kind of test. Do you want to augment that?

DR. KHOURY: No, I want to guard against genetic exceptionalism. So, I mean, I'm walking a very tight balance here. It is not only about clinical utility, but evaluating the whole spectrum from the analytic validity to the ethical issues.

The traditional medical model is clinical utility for coverage of new technology. But with genetic testing, I think we have to take a look at the whole spectrum of data and information, knowing that there are actually lots of gaps in our knowledge. If we really want to move forward integrating genomics and medicine, I think we need to develop all these processes to get that done.

We have spent a lot of resources and billions of dollars to map and sequence a genome. If we can't go the next steps in trying to figure out how it can be used in actual practice, I think it would be really not a good outcome for our country.

MS. BERRY: Debra?

DR. LEONARD: I think that with genetic testing, you are looking at an entire paradigm shift with the practice of medicine. In that you will have knowledge from testing one person that applies to an entire group of people, whoever is related to that person. With those other people instead of waiting for signs and symptoms to develop and then doing a diagnostic test and treating them, we will have the capability of potentially taking preventive strategies for those other family members, not having them wait until they have signs and symptoms.

So how you implement that is difficult. We are used to thinking of medical practice in terms of individuals and not in terms of families.

DR. TUCKSON: Let me just do this. Cindy has still got the helm here, but let's just do a reality check. We are at 12:30, we've got a guest at 1:00, and we've got to eat. We have a real cramped schedule here.

I think we heard just now one person describe what the problem is. We've heard Debra, she took a shot at it, and we didn't give her a chance yet to get into the level of details. She is talking now there is really a paradigm shift. That paradigm shift can be defined, I'm sure, by a set of characteristics.

Cindy, let me just come back to timelines. This is really important work, and it is important to get this one right. I still think we've got some more drilling down to do, particularly in defining the problem, and then being able to have a cogent set of recommendations, just for this section, much less a couple of other sections.

Cindy, when, again, I can't remember, is there a drop dead date by which this committee committed that it would have this report out? Is there a reason that this report has to be out the door on a date certain?

MS. BERRY: In the timeline that staff has suggested, the due date for additional edits after this meeting would be October the 29th. In November, the staff would prepare the next draft and the Federal Register request for public comments. The public comment period would be generally December to January, and then the next SACGHS meeting, of course, is February 28th to March 1st, where we would review the public comments received at that time, and finalize recommendations.

DR. TUCKSON: Well, it seems to me that we would like to keep that, at least to the last date, which is the March 1, you know, getting it out the door and into March is a reasonable timeline.

Now, I'm not sure, but what I think we will do is during the lunch period, I mean, clearly you as a committee are going to have to weigh in on some more of these issues. I don't think that the subcommittee itself can do it all without you being involved.

There is discussion that has to occur beyond the time that we have available right now. So we are going to have to figure out whether or not that means, again, through a conference call, or whether it means that we push everything back and bring more work back to this committee. I mean, we're going to have to figure out a set of tactics here.

But we can't resolve this issue right this second. I'm looking to you all for some guidance around how you want to move forward. We've got to decide this right now.

MS. CARR: Could I just mention a procedural issue? If the committee were to meet again in full committee through a conference call, we would need to make that available to the public. So you can meet in a closed session, a task group or a subgroup of the committee, but not the full committee.

DR. TUCKSON: Well, let me ask Cindy as Chair of this group, what is your thinking is on behalf of your committee? That is this. The one thing we don't want to do as a committee is extend every doggone report 20 cycles, and we never issue anything of importance, and we just look like an ineffective group.

Do you feel though that this will need to move back one meeting cycle so that we can come back, revisit, and tighten down on these issues that are unresolved? Or should we go ahead and try to get some stuff, some work done between the subcommittee and the full committee between now and the next scheduled meeting? Should we try to go ahead and keep to our schedule and just try to get it done outside of a formal meeting room?

MS. BERRY: I think the issues that are remaining that we haven't yet gotten to here probably can be disposed of pretty quickly. I think we can stick to the original time frame. The big kahuna of course is this big issue that I don't have a sense that we can really go back and draft anything, because there really isn't a consensus yet. So it may require a conference call of the full committee to do. I think by doing that, we still can stick to the original ? -

DR. TUCKSON: Can I ask the full committee, Debra, and part of your response, I know you all work real hard on this committee. I'm loath to ask you to do extra work, but would you be willing if we could structure a very tightly framed call that was very clear about the issues to be discussed, would you be willing to participate in that between now and the next meeting?

DR. LEONARD: Yes. That's the short answer to your question. I think given our earlier discussions on the genetic counseling issue, the data gathering that is going to happen, and informed discussions at the next meeting, I don't see any way that staff can generate a report to go out for public comment before our next meeting and gather those public comments. So I see no choice but to move back by one cycle, unless Sarah says you can work miracles.

MS. CARR: I think the committee has raised a number of really significant questions. I think we felt, staff did, that coming into this that there would be more. I know that I am somewhat diverting from what Cindy just said.

I think that this feeling I have is that you have raised some very fundamental questions that we need to gather more information for you about, and as Debra just said, a lot more information needs to be gathered about the counseling, the licensure issue, and so forth. So my inclination is to push it back one, even though I was raring to go and hoping this would be done by the next meeting.

DR. TUCKSON: Great. Cindy, would you accept the pushing it back?

MS. BERRY: Sure.

DR. TUCKSON: And then I think what we want to do is so that we don't lose the momentum, and we've got to really break off, I would urge the committee, Cindy, through staff -- Suzanne, Amanda, and everybody else -- let's try to get something back out to us right away that lays out where we are and what is uncertain so that while it is fresh in our minds, we can think about it. So that we don't lose the momentum of trying to drive this thing forward, let's start clarifying what really has to occur next. So we'll get that out to you right away.

Let me just close by saying to Cindy and the committee, you know, you've done us a great favor to get us this far. This is hard. So I don't think we should be disappointed.

To the staff, you worked your tails off on this thing, and we really want to thank you for that. We're going to keep at it. We'll get it done in short order.

With that, I think you are a very wise committee, and I think you've reached a wise judgment. Let's stop and grab lunch. We have a guest coming exactly at 1:00, so you need to be back in here around ten of. We'll have five minutes of conversation so we can justify the free food.

(Recess.)

AFTERNOON SESSION (1:00 p.m.)

DR. TUCKSON: We're back at the magical 1 o'clock hour. This train is running on time.

There is a wonderful opportunity to hear from the public. There is a person named Andy Faucett who is going to present. We are very pleased to have the perspective of the American Board of Genetic Counseling once again.

Thank you for your earlier contribution to the meeting, Andy. Please, you have five minutes. We're very interested in what you have to say.

MR. FAUCETT: Thank you, Chairman Tuckson, and other members of the committee. I really enjoyed the opportunity this morning to answer your questions. I'll just start by saying that please feel free at any point to use the ABGC as a resource. We really want to make ourselves available.

The American Board of Genetic Counseling is a national accrediting and credentialing body for the profession of genetic counseling. The ABGC establishes minimum requirements for graduate programs in genetic counseling, and develops a criteria by which individuals become eligible to sit for the certification exam.

ABGC also recognizes the importance of demonstrating a lifelong commitment to maintaining the knowledge and skills necessary to provide genetic services, and as a result, oversees recertification of genetic counselors.

ABGC feels that resolution of the two issues discussed by this committee during this meeting, genetic discrimination and building and reimbursement, are critical to the continuation and growth of the field of genetic counseling. One of the primary goals of the process of credentialing and accreditation provided by ABGC is to protect the public by ensuring access to professionals appropriately trained in genetics.

Comments from presentations yesterday and from working genetic counselors imply that some individuals are fearful of genetic discrimination, and are afraid to seek the help of trained genetic professionals. Such individuals may feel they must seek genetic information from other health care providers, non-medical care givers, friends and family members.

In requesting answers to important questions about their risks to develop medical conditions with an inherited component, they often receive incomplete or incorrect information. This could potentially result in an individual not obtaining information about optimal health care interventions and prevention programs.

ABGC-accredited training programs universally include the topic of discrimination in their curriculums, and teach genetic counseling students how to discuss the advantages and disadvantages of obtaining genetic information. Legislation designed to reduce

genetic discrimination and educational initiatives addressing the actual versus the procedural risk of genetic discrimination needs to be developed so that individuals may freely discuss their concerns about genetic conditions with professionals who can knowledgeably provide accurate information. The ABGC is willing to work with this committee and others to reach this goal.

ABGC also works to ensure that the field of genetic counseling remains a viable and attractive career. The difficulties with billing and reimbursement for genetic counseling services could impede the development of new genetic counseling programs, and interfere with the ability of institutions housing clinical genetics programs to support the activities of genetic counselors.

As discussed at the last meeting of this committee, efforts must be made to increase the number and size of training programs. University leaders will assess the viability of the profession, and the need for new programs and expansion of existing programs in genetic counseling before committing resources.

Lack of reimbursement for genetic services could result in a decrease in these services, affecting not only our patients and their families who are dependent on these services, but also decreasing the availability of clinical training sites for genetic counseling students.

Lastly, potential students may be hesitant to enter the field of genetic counseling because of the uncertainty of reimbursement for services. As this committee is well aware, the advances in genetics are forcing fundamental changes in the way health care providers practice medicine and think about health and disease.

Knowledge about genetics and its social and ethical implications is becoming increasingly essential for many health care professionals. Genetics health care professionals have been and will continue to be the ones who will train and educate other health care professionals about the many complexities of genetic medicine, including the potential for discrimination.

According to a professional status survey administered by the National Society of Genetic Counselors in 2002, a majority of genetic counselors are currently involved in the genetics education of physicians, medical students, and other health care professionals.

Many genetic counselors have developed and implemented innovative, educational models that facilitate the genetics education of other health care professionals and students. ABGC is committed to working with this committee to reduce the barriers of genetic discrimination and inadequate billing and reimbursement for genetic services, and encourage the public to seek information from trained genetic professionals.

As certified genetic professionals, we recognize the demand for genetic counseling services will continue to increase, and we would like to ensure that these services are provided by appropriately trained professionals.

ABGC supports this committee and its efforts to recognize those qualified to provide genetic counseling services, and hopes the committee will support the credentialing process already in place for genetic counselors, nurses in genetics, and others. Clinical genetics services must be recognized by the health care industry, and reimbursed appropriately, both so patients can receive quality genetic services, and genetic professionals can be trained.

There must be high standards for all genetics professionals, and competency must continue to be assured through accreditation of training programs and certification, and recertification of practicing genetic counselors.

Thank you.

DR. TUCKSON: Terrific. Thank you so much. As discussed earlier today, we really are looking forward to your report where you can sort of clarify the landscape for us around what is known and what is not known in the area of the certification and how that whole thing fits together with the reimbursement issue.

I think the more explicit, complete, and precise you can be, the better your efforts will be served, and the better our committee can do its job. So we really put a lot of faith in your report.

MR. FAUCETT: Thank you.

DR. LEONARD: Could you comment on the genetic counseling community view of how you see genetic counselors integrating into genomic medicine? Moving toward it being complex disease traits, and what implications that has for the workforce that would be needed, the genetic counseling workforce?

MR. FAUCETT: Well, I would have to say first that I don't think the community has reached a uniform decision about how that will move forward. But many of the models that have been talked about would use other individuals to help in the process. You may or may not have a genetic counselor who is looking at the more difficult cases or the more involved cases, kind of a triage-type effect.

But clearly the number of individuals that will be needed to help with those types of issues I think are very complex. My concern though is just because a test looks for five or six things at once and it would be complicated, that we would reduce the need for genetic counseling, where I actually think it might even be increased because of the complexity of the issues. Particularly if your real interest is to test for Disease 1, but they use tests for five diseases, and you have to find out about something you don't know much about.

So I think it is going to get more complex rather than less complex. But I do think we're going to have to look at models to involve multiple other individuals in the triage system.

DR. WINN-DEEN: I was wondering if you could comment on what impact it might have on the amount of time that a counselor spends with a patient if we actually had federal nondiscrimination legislation in effect and protecting individuals. Would this allow you to spend less time discussing those issues, and more time focusing on the real clinical issues?

MR. FAUCETT: Having done cancer counseling for a number of years, prenatal first, and then cancer, I would kind of agree with some of the comments yesterday, that the current section, about half of it is talking about the implications of the testing. A large part of that being the concern about how will it affect your health insurance, life insurance, and future medical care.

I think it could make a significant difference in the amount of time if you could be very reassuring to people that that was taken care of, and would make the process much more productive.

DR. TUCKSON: Any other questions?

(No response.)

DR. TUCKSON: Thank you very much, Andy. We really appreciate it.

MR. FAUCETT: Thank you.

DR. TUCKSON: Let's move on on the agenda. As you remember at our priority setting session in March, the committee identified gene patents and licensing policies

and practices as a high priority issue, because of its potential to effect access to genetic technologies.

We elected to defer in-depth consideration of the issue in light of the work that we became aware of being conducted by the National Academy of Sciences' Committee on Intellectual Property Rights in Genomics and Related Inventions.

The NAS committee began its work in February of this year. That study by the NAS is being sponsored by NIH, specifically NCI, NHGRI, NIGMS, NICHD, and the NIH Office of Technology Transfer. Tim Leshan, Director of the Policy and Program Analysis Branch at NHGRI, serves as the NIH liaison to our committee. We are very happy that he is here and attentive.

MR. LESHAN: Yes, very attentive. Thank you.

DR. TUCKSON: Now, we're very blessed that we have a guest from that committee, Dr. David Korn, who in addition to being my friend is a Senior Vice President for Biomedical Health Sciences Research at the Association of American Medical Colleges, and is a member of the NAS committee. He has kindly agreed to brief us about the committee's charge, scope, and progress to date. For your information, his biosketch is at Tab 1 of the briefing book.

I'll just say parenthetically here that the committee is at a very delicate stage in its discussions. So David will provide us with I'm sure absolutely the maximum amount of information that he can possibly provide us with at this moment. He may not be able to provide us with everything because of the status of his committee, but I'm sure he will clarify all of that as we go forward.

DR. KORN: Well, let me thank you all very much for the privilege of being here with you today to talk about this committee, which I had a role in instigating, I think is the fair thing to say.

But I also have to tell you that the National Academy has very ironclad rules about what can and cannot be said about any committee that's underway. So it is sort of like I can give you my name, rank, and serial number, sir, and that's about it. But no, it can be better than that. I really cannot even hint at presenting to you any flavor of discussions or conjecture about possible findings and recommendations of the committee. It is simply verboten. So I can't do that, and I apologize.

But in any event, this committee was put together, and the members of the committee are shown on this slide. I think it is just worth noting, Shirley Tilghman, who is a very eminent developmental biologist in her past life is now the President of Princeton University. She co-chairs this committee with Rod McKelvie, who is very well known in the patent law business, a former judge in the courts in Delaware, in which more companies are incorporated than any other state in the United States.

So Delaware courts have a very rich history in dealing with corporate issues, including intellectual property. Ashish Arora is a Professor of Economics and Public Policy at Carnegie Mellon, who with others, including Wes Cohen at Duke, and such, has spent a lot of his recent career studying the economics of innovation, which certainly patents relate to.

Helen Berman is a Professor of Chemistry and Chemical Biology at Rutgers, and is the curator of probably the most definitive web-based repository of protein crystal structures, and cares much about proteomics research. Joyce Brinton has been, and is

retiring now, the Director of Technology Transfer at Harvard University.

Stephen Burley, a former academic, I believe at U.C. San Diego is now the Chief Scientific Officer at a small biotech company. Todd Dickinson is now the Intellectual Property Counsel of General Electric. Under the Clinton administration, he was Commissioner of the U.S. Patent and Trademark Office.

Rochelle Dreyfuss at NYU and Rebecca Eisenberg at the University of Michigan are both eminent scholars, Professors of Law in the area of intellectual property law. Very well known scholars. Charles Hartman is an investor who is into starting up biotech companies.

Dan Kevles is a very well known historian of science. He spent a large part of his career at California Institute of Technology, and more recently was recruited away by Yale University. One of his better known and more recent works concerns the David Baltimore case that some of you may have been familiar with.

George Milne is a former Senior Corporate Officer of Pfizer, now dabbling in investments and biotech start ups. Richard Scheller is a former faculty member of mine at Stanford who is now the Executive V.P. for Research at Genentech.

Rochelle Seide is a litigator. She is in the private practice of law and specializes in intellectual property law. Bob Waterston's name is known I think to all of you. One of the key contributors to the human genome map, now out in Seattle.

Nancy Wexler is well known to all of you for her work in hereditary disease, especially Huntington's disease. Brian Wright is a Professor of Agriculture in Resource Economics at U.C. Berkeley. It is a very strong department at Berkeley.

So what we were asked to do by the NIH was to examine trends in the number and nature, well, you can read this. But it is basically what kind of patents are being issued and granted to technologies related to genomics and proteomics.

Some study of the procedures that our patent and trademark office is using are relative to those used by others, specifically in Europe and Japan. How the patenting of genomic and proteomic inventions and/or licensing practices may be affecting research and intervention and, based on our findings, recommend steps the NIH and others might take to ensure the productivity of research, innovation, and so forth.

DR. LEONARD: David, could you clarify? I thought part of the charge was also to look at clinical impact, but I'm not seeing that on your list.

DR. KORN: The charge slide that was given to me by the committee did not list it, but I will come to it, because that is a topic that the committee has been looking into.

I just wanted to point out to you that there is a website that has all of the documents and agenda of all the meetings and almost anything you want to know about this committee, you can find on that website.

So what sort of things have we looked at to date? One has been the policies, procedures, and operations of the patent in the trademark office. That focuses of course on the criteria which are established in U.S. law, and about any one of which we could probably spend half a day talking and arguing, because every one of these criteria has been challenged in the area especially of gene patents. That is patents that have been issued, and have been challenged on the basis of their utility, and on the basis of their novelty and non-obviousness. That means to a person skilled in the field. On their written description and enablement, especially vis-a-vis the scope of their claims.

That is you claim something when you file an application, and you are supposed to demonstrate convincingly that you have what you are claiming, and that you have actually got it. Not you are thinking about it or wishing about it, but that you actually have it. There has been some quite heated discussion in the literature about patents regarding the perceived variance between the scope of some claims and the written description and ability of backing them up.

Then we are going to be looking at international policies and practices, Europe and Japan. We are looking for evidence about ways that individuals, entities, and companies have managed what Rebecca Eisenberg has dubbed the "patent thicket." Patent thicket means you want to do something. In order to do it, you find that you have to negotiate intellectual property rights with a dozen, two dozen, or eight dozen different owners of bits and pieces of what you want to do.

A very interesting example of a case where that has recently occurred is in golden rice, which was developed for the specific purposes of dealing with Vitamin A deficiency. In order to get from the discovery the invention of golden rice, which was a very difficult and long-time genetic technology problem in ag biotech, they had to work their way through about 40 or 60, I think, patents on different bits and pieces of what they had done to get the product.

All of that had to be negotiated before the discovery could be then reduced into benefit for people. That is, before you can get a company to begin manufacturing it, all of this intellectual property stuff had to be negotiated. I gathered from a paper in Science last fall that it took well over a year to do that.

One of the reasons it succeeded was because nobody thought anybody was going to make much money on this. It's going to be aimed mainly at a developing world where people need this foodstuff, but there certainly isn't a lot of money.

There has been a fair amount of discussion about the research exemption. Does it exist? I'm going to divert, if I may, for a couple of seconds or minutes, because I'm very interested in this for obvious reasons.

There is a case that was decided by the appellate court in the United States that deals with intellectual property. It is called the Court of Appeals for the Federal Circuit, or the CAFC. Unlike all other jurisdiction in this country, there is only one appellate court that deals with all intellectual property cases that come out of district courts. One appellate court. It basically makes the law on patents. Rarely, rarely does a case get bumped up to the Supreme Court from this court. That is, the Supreme Court shows this court great deference.

So what is the issue? Well, the issue is that after the patent statute the Constitution defines the rights of artists and inventors to have exclusive use of their inventions for a period of time to promote progress in the arts and technologies, I think it says.

But in 1813, a Justice, in a case known as Whittemore v. Cutter, wrote in an opinion about a patent infringement these words, that "It could never have been the intention of the legislature to punish a man, who constructed...a [patented] machine merely for philosophical experiments, or for the purpose of ascertaining the sufficiency of the machine to produce its desired effects." That was the first legal writing that said it might be possible to "infringe" a patent and not really be infringing, yet in terms of the legal sense that you could do things with it and not be infringing.

The same Justice, in the same year in another case some months later, went

on to say that, "Patent infringement must concern the making of the patented thing with the intent to use for profit, and not for the mere purpose of philosophical experimentation, or to ascertain the verity and exactness of the specification."

So working on the patent to understand it better, working on the patent not for profit, but for philosophical experimentation, he said were allowed. For almost 200 years, that has essentially been the base of case law. That means this is not in statute, but it has been accepted as case law by other judges.

Why is it important? Because at least in universities where a lot of this research that you're concerned with gets done, it has generally been assumed that they are operating a non-commercial, not for profit, seeking knowledge, and understanding fashion, and they have been cloaked, protected, by this experimental use exemption.

Well, along came a professor named John Madey, a physical scientist of some accomplishment, who sued Duke University in a case that went up to the appellate court over the use of some form of free-electron laser equipment on which he held patents. He had been a major inventor of free-electron lasers.

This court now in 2002 said that "Any act in the furtherance of the alleged infringer's legitimate business and not solely for amusement, to satisfy idle curiosity, or for strictly philosophical inquiry, that act does not qualify for the very narrow and strictly limited experimental use defense."

Now, I don't know whether Hunt Willard and others would say that what they do every day in their university work is to satisfy idle curiosity, or for amusement, or for strictly philosophical inquiry. But the court then went on to say, "Our precedent does not immunize use that is in any way commercial...[nor] any conduct in keeping with the alleged infringer's legitimate business, regardless of commercial implications."

So for the first time now in the law since 1813, the idea that you could do these things if it was not commercial has now been blown away by this CAFC holding. It went on just to rub the salt in a little bit more, that "Such activities as obtaining research grants and educating students and illuminating faculty unmistakably further the institution's [Duke's] legitimate business objectives."

Now, given that, I think the answer to whether or not there is a research exemption at this moment is highly in doubt. This has some relevance to the point that I think Debra Leonard wants me to get to, which I will. We are trying with other organizations here in town and the AAAS to do a blinded survey of university experience post-Madey to see whether there are indeed challenges and demands for licenses and payments, and whether he used various kinds of patented inventions that are widely used as research tools, widely used, and almost anybody working in current genomics and proteomics is using somebody's research tools one way or another.

There is also some effort to draft a statutory exemption for research, such as that by the American Intellectual Property Lawyers Association. But I won't take your time with that. Their proposed use, their proposed exemption is exceedingly narrow, and would not cover most of the kinds of research uses that university faculty and businesses doing research, I mean, basic research and not necessarily product development, would be protected by.

We have been looking at the issue of licenses, as well as patents, and material transfer agreements, which are these contractual documents that faculty use when they are sharing materials, reagents, organisms, cell lines, whatever, with others either in industry or

in universities. These things have become a major burdensome impediment to the sharing of research reagents and tools, and living organisms.

This is not the patent law, these are material transfer agreements. Then at the last meeting, they did turn their attention to the effect of patents on genetic testing and genetic gene patents on the practice of medicine.

The issue here is this. That there are, as you may know, a number of cases now where a genetic mutation has been described that has let's say some kind of correlation with an interesting phenotype. It may in fact be the dominant gene, or it may be a gene with some statistical correlate, and one doesn't really know what its pathophysiological role is.

The patentor of the gene can essentially obtain a patent, and the patentor of the mutation can essentially obtain a patent that will prevent anyone from using that patent without that owner's permission. What has happened in some of these cases, such as Canavan's and one of the hemochromatosis genes, and I know Debbie can give you others, is that the owner, which is frequently a university, by the way, almost always a university, gives a highly restrictive license to A, the provider of the test.

Let's take BRCA. The provider is Myriad Genetics. They have a worldwide exclusive on the practice of the test. The fact is that if they choose to enforce their license, which they do, they will say that no one in the world has the right to do a BRCA test on any specimen, except them. It is a nice business position to be in, I have to say.

By the way, let me make clear now that I am expressing my personal views about this. I think I'm being factual about what I'm describing, but the views are mine and not the committee's. I don't want any misunderstanding about that.

So the issue that I think people like Debra Leonard and others who are in her field of medicine are concerned about is that these patent owners and licensees are not intending to develop their patent by providing test kits, special equipment, special instruments, or special reagents that other users would probably eagerly buy if they were of high quality and reliable. They are simply saying nobody else can do it.

There is an argument made here that this essentially is saying that an owner of a piece of knowledge, a quantum of information, a bit of information, can essentially prevent other qualified professionals, physicians in this case, from using that knowledge to deal with their patients. They have to go to the exclusive licensee in order to get the work done that will tell them what is going on with a given patient.

This is the issue that is embodied in the words that are here on this slide. Now, I will tell you that in our country, there are two statutory exemptions. Statutory, that is in law, passed by Congress through the Patent Act. One of them in 1984 known by its authors, Senator Hatch and Congressman Waxman, allows activities and uses reasonably related solely to developing information required to secure FDA approval.

What this is really about is generics and allowing those who want to make generic copies of patented products to begin to develop the information on their generic that they need in order to go to the FDA and get approval to sell that generic when the patent term has expired.

They don't have to wait until the patent term expires to begin all of that preparatory work. They gain some years by being able to start working on it ahead of time, and having their package ready to give to the FDA.

That exemption has also been reduced by this appellate court in another

recent case. They clearly don't like exemptions, this court. But it does exist.

Now, the other one, which is more pertinent to Debra Leonard and her colleague's issue, is the First-Ganske, Senator First and former Congressman Ganske, amendment of 1994 which permits medical practitioners to practice patented medical and surgical procedures.

What is a patented medical and surgical procedure? Well, in this instance, it was an ophthalmologist who wanted to patent a type of curved incision in the cornea. He wanted to own it, patent it, and control its use. There was a lot of unhappiness about that sort of thing. So this amendment was passed to physicians First and Ganske, patented medical and surgical procedures on a body. That's your body, my body, a body, but excludes the practices of processes that would violate biotech patents, and specifically the provision of clinical laboratory services regulated under the Clinical Laboratories Improvement Amendments, which are known as CLIA, which is the statutory body of law that basically regulates diagnostic laboratory services performed in the practice of medicine.

These are the only two statutory exemptions, as I say, and the First-Ganske explicitly excludes laboratory diagnostics which would be the kind of medical practice that we are talking about here with use of mutations in diagnosis. So they are explicitly excluded.

Just to finish, the committee is on a very fast pace considering the complexity of this topic, and expects to be finished at an April meeting, and release a report by June, which I think will be an extreme amount of work that has to be done to get to a report. But that is the schedule the committee is on.

I think that is what the committee is up to. Those are the topics that it has looked at. I think I'll stop here. I'd be happy to take any questions that the Chair permits.

DR. TUCKSON: The floor is open. Ed?

DR. McCABE: Dr. Korn, you mentioned the Canavan's example, and you talked about exclusive licensing. But Canavan's takes that one further, where it is exclusive, an attempt at exclusivity that basically removes a diagnostic from patient availability. Can you comment on that, please?

DR. KORN: Well, let me try, Dr. McCabe, and see if I do it addressing your issue.

The argument is raised that if a particular, and this is all David Korn, and not the National Academy and its committee, okay? It is my opinion. The argument has been that when there is an exclusive provider of a laboratory diagnostic test, one, they can set whatever price they want, in that is there is no competition. Two, there may be an inconvenience in getting samples from wherever the patients are to wherever the privileged provider may be. Three, that there is no real independent check on the validity of the work being performed by that provider.

That is if you get a result, you cannot go to another licensed, credible laboratory and have that result checked. Sometimes results are wrong. If you don't get an independent check, you can have some really unfortunate consequences that were not necessary because you are proceeding on a problem that may not exist, or a problem that is different from what you were told.

All of these are arguments about the undesirability of having an exclusive provider of a diagnostic test, and you probably know if you read the newspapers that in France, and I believe in Canada, the government is actually backing professionals who are challenging

the Myriad patent on BRCA1. They are doing it on the grounds of protecting the public health of their particular societies. That is an answer.

Now, I'm a pathologist. At least I used to be. To my knowledge, there has never been in the practice of pathology, and Debra can correct me, any such exclusivity imposed on the practicing credentialed community. There certainly have been instances where practitioners will choose to send specimens to reference laboratories, because there may be very rare diseases, or they may be very difficult tests that require a lot of special expertise and equipment.

It is often an economic, efficiency, and a quality decision that says, it would be better to send it over there to that lab because they do them all, and they know what they're doing, rather than have our lab try to do something that we may see once or twice a year or two.

So certainly there are reference labs that do have semi-monopolies on certain tests, but that is certainly by the desire of the professionals that it is the best way to deliver the care. It is not imposed. There certainly are companies that have been very successful making important diagnostic laboratory equipment, like FAC sorters, robot chemists, and all. They have a right to protect their patents and make sure you don't try to build your own and copy their machine, and then use it for your own practice.

But most people who aren't nuts would be happy to buy good equipment, good reagents, and good kits, because first of all, they are reliable. Secondly, it is easier, it is simpler, it is easier to teach the technicians how to do it. So there is a great desire to have these kinds of things, and people will buy them and use them if they are available.

But in these instances, that is not what we're talking about. It is not a product that somebody is trying to use illicitly. It is a piece of information. That is what is different.

It worries me about what it means for the future medical practice, frankly, because we are still at the dawn of the genomic revolution. We are still at the dawn. I mean, there will be gazillions more sequences that are going to have some kind of relationship with various human traits and disorders of interest.

The idea that every single of them might be restricted from use in this way, I just find a very distasteful scenario. It just seems to me a path that medicine has not taken before. I don't see it has a happy path. But that's my personal opinion.

DR. TUCKSON: Emily is next, and then we'll just go right on around like that.

DR. WINN-DEEN: So I just wanted to ask you if you could say a little bit more than you're going to generate a report. Can you talk about at least sort of maybe the table of contents? Is this going to be just a review of the state of the state? Or do you intend to make some specific recommendations? You don't have to say what the recommendations are, because I understand they --

DR. KORN: I think the committee is fully empowered to make recommendations. I don't know what kind of recommendations that it will choose to make. I do believe that there is a wealth of perspective, expertise, and experience from the roster. It is a very distinguished group of people. I expect that they will make findings, and they will make recommendations.

DR. TUCKSON: That was good.

(Laughter.)

DR. TUCKSON: Brad?

MR. MARGUS: So can you help me again with the distinction with genetic information? It seems that any patent gives the inventor a monopoly, and that can be troubling, but that is the way our system is, and it is necessary for a lot of reasons.

So I'm trying to hear. It seems like the explanation in this case is that it is different because somehow human health is involved, and there is more risks that it may not be made available to everyone.

I understand that, except drawing the distinction between genetic information and any drug for which there is a monopoly, and there is the risk that Pfizer or Merck will not sell their drug to everyone in the world, or make it available to everyone, just in the same way as the genetic test wouldn't be available, and the drug may not be available. Yet no one is suggesting that we don't ? - the drugs.

If the difference is that there is a pill instead of a piece of information, can you elaborate on that? I mean, in either case it could have taken 20 years to develop that piece of information, just as it takes 20 years to develop a drug.

DR. KORN: Well, you know, as a matter of fact, a lot of times information is available to those using the information well before a patent is ever issued, as it turns out. But that's not the point.

I think the issue is that historically at least, the practice of medicine has not been limited in terms of what practitioners may know and apply to their patients, or do to their bodies, because somebody owns something. I mean, if you look back at the history of medicine, it is very economic. That is, people got their goodies, their kicks, but when your name is on things, you know, you have a lot of surgical procedures that have great surgeon's names associated with them.

Jones' approach to the gall bladder, and Smith's approach to the left kidney, and this, that, and the other thing. They got their pleasure by describing these approaches, and then people use their name to describe it. They didn't say, I'm going to be the only one who can do this, or I and these two people that I'm appointing are the only two people in the world who are able to do it. They let the professionals use the information.

I think that is a tradition in medicine that I have great respect for. I just find it offensive, personally, to be told that this is a piece of information that you may use in your practice, and you're not allowed to. I mean, that doesn't mean it is wrong. It is certainly legal, it is certainly legal. I just find it very distasteful.

I have trouble envisioning medicine in 2050 when there are, as I said, gazillions of these bits and pieces and quantum of information that all have intellectual property tied onto them. Now, you could say to me, that's just tough cookies. You know, you are living in a new age --

MR. MARGUS: Most of (inaudible) today will be (inaudible) by then.

DR. KORN: Well, maybe. But the point of the matter is, and I would certainly defer to your expertise on this, I imagine that if you are the sole repository of the mutations in a particular gene of interest, that you can always selectively file for more patents on a different mutation in the same gene that might have equally interesting clinical correlates, and kind of keep rolling your ownership of the gene out forever and ever and ever.

I don't know. Is that possible?

DR. LEONARD: Brad, can I distinguish between drugs, which have patent

protection, and the gene sequences, which also have patent protection?

With a drug, you are patenting a chemical compound that reacts in some way in the body to prevent or treat a disease, or change how something works. You are not patenting how that something in the body works. So anyone who wants to create a different chemical to treat that same disease can do so. They just can't create the same chemical, and produce and make it.

With gene patents, you are really patenting. Some of these patent the sequence any way whatsoever you can imagine under the sun of looking at that sequence, or detecting in someone's body, or in a specimen, or anything. So it is like patenting the disease rather than the chemical compound that treats that disease. There is really a fundamental difference between drug patents and these gene patents.

DR. TUCKSON: Thank you for that clarification.

Hunt?

DR. WILLARD: Just before I get to David on that point, of course there are patents. It depends on how sweeping the claims are. There are claims that are broad enough to capture an entire biochemical pathway, regardless of the particular drug.

But to David, is your committee, if you can say, also looking at the concept of defensive patents and depositing rights into sort of a scientific commons for every man's use? Or is that something that is off line from what you're looking at?

DR. KORN: I believe that they certainly are going to have ? - there has been conversation about defensive patents. Certainly they are aware, very aware of the initiatives like the Bermuda rules on the human genome sequence, and the SNP Consortium which is an industry, largely an industry consortium. The HAPMAP project, they are very aware, and they are aware of Helen Berman's protein structure bank.

Please be aware that if there is an issue here worth your attention, that it is not business versus academic or commerce versus university, because I believe that the largest holder of human genome sequence patents in the United States is the University of California System.

So this is not universities are good guys and businesses are bad guys. Not at all. This has nothing to do with whether you are a university or a start up, or a major pharma company. It is an issue of, well, to me it is an issue of is it in the interest of the health of the public to proceed along this kind of pathway or not?

I think you all have plenty of capacity to debate that problem. As I say, France and Canada at least have decided it is not in the interest of the health of their people to recognize such a sweeping, exclusive patent.

DR. TUCKSON: We've got three people in line. One is the aforementioned person from the University of California System, Ed McCabe. Then we have Debra, and then Tim.

DR. McCABE: Well, I take full responsibility for everything that occurs in the University of California System.

I was following up. You talked about some of the different issues with exclusive licensure. One of the areas that the American College of Medical Genetics discussed in their statement also had to do with education.

If there are patents on methods or processes that are exclusive, then it is hard to educate the next generation of young people regarding how to utilize those. By the

time they go off patent, we could lose the expertise.

I'm not asking for a conclusion, I'm just wondering if the committee is taking this up.

DR. KORN: I don't know the answer to that, or I can't remember whether it has come up or not. Debra Leonard presented to the committee at its most recent meeting a few weeks ago. I don't remember whether you mentioned training.

DR. LEONARD: David, is there any way that this committee could help your committee, is there anything that we can do to inform the committee or, you know, anything?

DR. KORN: It seems to me that your charge is the health, genetics health in society, isn't it? It seems to me that you might, if you choose, to put more energy into the issue of how these sorts of intellectual property rights should be managed to make sure that the health of the public, which is your responsibility, is maximized, or certainly not impaired.

How you would do that and what recommendation you might make about it, I don't know. It seems to me that that would be very squarely within your charge. As I say, I'm a capitalist, you know, I mean, I'm a product of a capitalist society. I'm not really trying to undermine our economy, as I have been accused of doing by some.

I do think it is a genuine issue here of what is best for the future health of the public. If this is the best way to do it, then that's fine. If it isn't, then maybe there needs to be some modification.

DR. TUCKSON: Just as an amendment, David, to the answer you gave Debra. Do you think from your sense of what your committee is going to produce, that our attending to the charge you just sort of recommended for us should wait before we look at that until your committee finishes its work?

DR. KORN: I think as a Secretary's advisory committee, you are free to do whatever you like, when you wish.

DR. TUCKSON: Let me rephrase that.

(Laughter.)

DR. TUCKSON: We love that freedom. I guess I'm just sort of wondering, from your sense, do you think that your committee will introduce information that will be useful to us in undertaking that?

DR. KORN: I don't think I can answer that. I don't know.

DR. TUCKSON: Thank you.

Tim?

MR. LESHAN: I just wanted to say as the liaison from the NIH, thank you, Dr. Korn, and the committee for the work that you are doing on this. It is such an important issue to everything that we are doing at the Genome Institute, and at the NIH in general.

I wondered if you might talk a little bit about some of the difficulty of gathering data to assess the impact that patents are having on genetics, genomics, and proteomics research.

DR. KORN: Yes, thank you for that, Tim.

It is very hard to do this. Apparently, I mean, what you are dealing with are anecdotes rather than data in many instances. That is why I mentioned the golden rice example as one that is not an anecdote, because it was actually described in a Science magazine policy forum a year ago.

Any national academy committee feels it important, and it is very understandably so, that their recommendations and conclusions, conclusions and recommendations, be data supported. Supported by data, driven by data, not by hearsay. Any academy committee.

I think that the committee is very interested in obtaining good, credible data that would shed light on some of these hypothetical concerns. It seems to be, this is not my field, of course, but it seems to be very, very hard to do so.

Certainly within the timeline of the committee's remit, it is certainly not possible to do a thorough three-year study, or something of this sort, to canvas the universe. But they are very interested in obtaining data that would help them to understand, and then think about what they might recommend on these issues.

I will offer another personal observation, however. Many industries have, and I wish there were a good patent lawyer in the room. But many industries have come to the desirability of patent pooling, of developing mechanisms to share their patents so that the whole process of development doesn't get swamped in a mire that nobody can move because everybody owns a bit of what has to be done.

The idea of patent pooling, to my knowledge, in biotechnology or biomedical research and biotechnology, hasn't yet existed. It is not something that comes easy when you talk about biomedical and biotechnology research. For example, I'm told by the experts that patent pooling were essential to the development of aircraft radio, certainly more recently in our lifetimes, in semiconductors. I mean, there is a whole tradition in semiconductors of companies who have bitter competitors, sharing research investments and knowledge at a very fundamental level of the science. I know that.

But in biomedicine, the early capture of discovery, very early capture in the pathway from discovery to some product, the early capture seems to be the name of the game. Of course you have whole biotech companies founded on bits of knowledge. Really early stuff. So there is a tradition of not having a lot of patent pooling.

It may be that in time, if things get mired down enough, that patent pooling will appear to be a very mutually beneficial way for everybody to go, where they put all their patents in a pool on agreeable ground rules and people can use that patent pool to move the field further on.

It doesn't, to my knowledge, and I may be wrong, it just doesn't seem to be something that comes natural to our particular area of work.

DR. TUCKSON: Well, we are right on schedule. But I want the committee to make sure that if we take ? - if there is one further burning question, to take good advantage of David Korn.

MR. MARGUS: I need to make a comment. Just so you know, today is my last day on this committee, so I've been dying to make a comment that would be a little more inflammatory. So here it goes.

(Laughter.)

MR. MARGUS: I'll preface it by reminding everyone that I'm extremely conflicted and biased in this comment. I'm conflicted because first, I run a company that isn't a gene-finding business. I'm also conflicted because I spent a lot of years trying to find a gene for my kid's disease that was an obscure, rare disease, and it was really hard to find anybody that would work on it.

So for all those reasons, I guess the only thing I would like to encourage the patent process reviewing committee, is that to not forget that the race to find the genes could really be important. Even if it seems like we're sometimes overwhelmed and flooded with so much information and we've got enough to work on for now, I think that continuing to encourage those gene discoveries, and there are a lot more to be discovered, is critical to accelerating and focusing biomedical research.

So I absolutely agree that if someone came along who found a replicated, true association between polymorphism and a phenotype of importance and they didn't make a test available to the world so people were being deprived, and maybe the government should have some march-in rights, or maybe the patent line should be made shorter.

But at the same time, you know, from the biotech industry, as you know, there is a need to find investors. Those investors only fund expensive projects to find genes if there is some kind of exclusivity or clinical monopoly that's possible. That's the hot button for every investor.

So on the one hand, we want to protect people from not having an important test, but on the other hand, I would hate to hear that one was done where suddenly there is not enough interest in continuing to find genes. I know the NIH has a lot of funding, but does not have enough funding for all the work that could be done in genetics.

DR. KORN: I absolutely share your view. I certainly would wish to do nothing ever myself that would shut down incentive to find genes. But I just would say there are ways that that incentive could remain, and there still could be more freedom of applicability of the discovery through compulsory licensing schemes.

I'm not recommending these, but they are just things that when he has talked about compulsory licensing schemes, the royalties that are not so punitive that nobody in their right mind could possibly make it work, or other things like that.

I mean, I think there is a middle ground, and I just don't know whether total exclusivity ? - by the way, I want to raise one thing. The recent Canavan decision in the Florida Federal Court that said that the families who had done so much to isolate the Canavan gene had no rights really in any of the resulting patent revenues and so forth, has led other groups, such as the pseudoxanthoma elasticum, PXE, group to form their own way of dealing with that, where they have created a not for profit foundation, I guess.

They are going to control the goods, the specimens, the tissues, whatever. They are going to hold the intellectual property, and they are going to have a voice. What happens if genes are discovered that are of use clinically? They are not discouraging invention, but they are going to have a say in how the fruits of the inventions are distributed among the people.

That's not a bad idea, that's not a bad way to go. There are other such diseases groups that have formed similar controlled mechanisms to keep a hand on what happens to the stuff that they are promoting.

DR. LEONARD: And given the coverage and reimbursement issue that we were discussing earlier for genetic testing, it is so inadequate that I can't believe that it is a really strong business model for a lot of gene diseases or genetic diseases. Rather, I think that your investors would be more interested in what drugs you could develop, or even test kits and other things that have true market potential.

MR. MARGUS: So when we discuss clinical theranostics in the next

session, pharmacogenomics, we'll hear how they are all tied together.

DR. TUCKSON: Well, let me thank you all for this.

Brad, you'll have further opportunities in the coming minutes to be provocative.

(Laughter.)

DR. TUCKSON: But let me thank David Korn, who did a terrific job. Thank you, David.

David, would you remind us again? Your report, I couldn't remember the date in which you think this thing is going to be submitted. I think it was June? June, 2005.

DR. KORN: June.

May I do one of those sales pitches? In December of 2002, we (inaudible) special issue of academic medicine that really presents a very new bias (inaudible) gene patenting issues. I know we gave Sarah a bunch of --

DR. TUCKSON: And we passed it. The committee actually has gotten a copy of it.

DR. KORN: If anybody wants more, let me know.

DR. TUCKSON: Okay, David. Thank you.

Let me, as far as this issue is concerned on our plate of activities, can I get a general consensus from the committee that we will revisit this issue upon the completion of the NAS committee report in June? And that we feel like they are moving forward, although, you know, we've got a lot to see in terms of what actually comes out.

But you can't find a more stellar committee working more assiduously and rapidly to get a conclusion done. So I think we would be well served by waiting for that. Does anyone disagree with that?

(No response.)

DR. TUCKSON: Then that's terrific. Let's move onto the discussion of our proposed plans for dealing with other priority issues. Let's start with large population studies.

Hunt Willard?

DR. WILLARD: Thank you, Reed.

I'm supposed to lead the discussion for us as a committee to plan how we wish to move forward, or not move forward, on planning a session or meeting for large population studies.

The task force that was to deal with this issue was a dream task force for those who always cringe at the thought of being appointed to such things, in that we actually neither met nor even were able to schedule a telephone conference. So we all worked indirectly by email and returns of faxes to Amanda and Sarah and the team.

What you have in front of you is a three-pager setting out from the staff perspective with input from members of the task force on potential topics, and I stress the word potential, for areas that we might productively spend some time focusing on.

I think my advice to this conversation here would be to briefly discuss the pros and cons of the different potential topics, and then move into a discussion, a reality-based discussion, of how actually we would fit this into this committee's game plan moving forward.

It is very tempting to conclude that everything is a top priority, and yet, we're backlogged in our future meetings. Certainly I have great enthusiasm for this topic as being an important one that this committee could weigh in on, probably needs to weigh in on,

and that the Secretary needs to be informed of. But I'm also aware that there are other groups within HHS that take this issue equally seriously.

So in front of you are four, really three different topics, and then the fourth one is sort of an all purpose wrap up. Four potential topics dealing with allowing this committee to become more informed about the nature of cohort studies that are under way in other countries around the globe, projects that are either underway, or under discussion in this country, and then the inevitable but certainly important social, policy, and ethical issues that are relevant to the planning of a cohort study in this country.

I think to me, it is just trying to frame the issue. There really are two questions. But both of them really are around the issue of should this country, should we advise the Secretary to support finding resources to mount a large cohort study in this country? What information do we need in order to make that assessment?

The United States is organized differently, both as a society and certainly as a health system, than other countries around the globe, and it may be that there are both some advantages as well as disadvantages to trying to mount an effort like that in this country that we would need to debate, and become informed in doing so by learning more about efforts in other countries where there are different organizational structures, both in terms of society and their health systems that have enabled them to do what they are doing.

The suggested names that are on this list in front of you are nothing more than a sort of menu of possible individuals who might be called upon. It is not an exhaustive list by any means. So I would urge the committee not to take these as being prescriptive in any way, or limiting in any way, but simply potential opportunities.

So Mr. Chairman, I would just open it up to committee discussion. We can either take it topic by topic or just general comments on this particular issue.

DR. TUCKSON: Before we actually do that, can I ask one background question? I think I'm also confused about where is the state-of-the-art status now within HHS around this population studies issue? Because I know that somewhere along the line, somebody is kicking around, and I think it is actually, Alan, in your area. And Sherrie Hans of the VA also may know something.

So would you mind, Hunt, if I could just get the background as to where they are today?

DR. GUTTMACHER: Yes, that's a very good question. Thank you. I think the way you put it is interesting, and probably an accurate description, being kicked around within HHS. I think that is probably the technical term of art for this.

There is a group within the NIH that is trying to figure out what the science of such a study might look like. A working group was empaneled to explore this topic over the course of the summer. They explored it. Staff of a couple of NIH institutes are looking at this and trying to develop a document, or at least some sort of summary of the science of this for higher ups at the NIH, and potentially the department to take a look at.

So that is where we are at the moment, is trying to figure out exactly what the science of such a study would look like, what the pros and cons of different participant sizes might look like, something about of course what the costs would be, and what kind of phenotypic, genotypic, and environmental exposure data one might want to gather.

DR. TUCKSON: Could I assume that some of the people in HHS that are looking at this would include at a minimum, NIH and CDC?

DR. GUTTMACHER: CDC and other agencies have been somewhat involved through the working group. But this is really primarily at this point an effort within NIH to figure out the science of that with some communication with other agencies for their expertise.

DR. HANS: Really, the VA is in sort of a similar spot. Perhaps even earlier than what Alan just described. We are sort of kicking around the idea as well. We've taken a little step back, and are spending some time talking to the communities that would be involved, both internal and our external constituents about this. So it is very early on in the discussion of what we might be interested in.

DR. TUCKSON: And finally, do we have any idea of, or do you all have any timeline by which you want to bring your fact finding effort to closure? Or is this an open-ended process?

DR. GUTTMACHER: I sure hope it's not open-ended. We would certainly aspire to have something more that we could share with the committee, for instance, at its next meeting. That would be our hope. Whether that will be realistic, I'm not sure, both in terms of trying to get the science of this gathered, but also depending upon what that says in terms of people higher up within the department or other parts of the federal government, feeling that it was ready to bring something up for comment. I don't know whether that can be realized by February or not, but that would be my hope.

DR. TUCKSON: And finally, given the door that Hunt opened up here, is there anything that we can do, that we need to do, to facilitate you all's efforts? Do we need to ? - I'm just making sure that it is not something as easy and simple as do we send a letter to the Secretary saying that the VA and CDC and NIH genome folk all ought all to get on the same page, or are you all capable of doing that without spurring ? -

DR. GUTTMACHER: I think we're pretty capable of doing that without being spurred, particularly because I think the realization of all those involved in this that if the federal government were in any way to participate in such a large study, it would require both the expertise, the logistical support, and the finances of more than any one of those agencies.

So I think even if those partners didn't want to play together, which I believe they do, they would have to play together to achieve this. So I don't think that needs to be ? - on the other hand, I think it might be helpful if the committee felt that that was appropriate for them, to recommend to the Secretary that such a large cohort study has real potential to be of help, and you would recommend that the department take seriously the question of exploring such a thing, and that it has potential real benefit to the health and well being of the American public.

DR. TUCKSON: By the way, I really appreciate the way you phrased and answered that. But I think that what you have said, and I want to just be explicit, is that the work of determining whether there is something there and can be done, is really going forward.

DR. GUTTMACHER: Yes.

DR. TUCKSON: And so you really don't need us to be bugging anybody about it right now.

DR. GUTTMACHER: I don't think you need to bug us, but I think it is helpful for a group outside of those doing this work who have expertise to say to the Secretary, as your advisory committee about genetics health and society, do you realize that a large population-based cohort study prospectively done could bring real value if that is something

that the committee, a statement that the committee would be willing to make. I think having the Secretary hear that from the committee could be quite useful.

DR. McCABE: I would see it a little bit differently, because I think there probably are going to be competing influences, or competing projects within HHS. You know, and so that I would be cautious that we not find ourselves saying ? - maybe we could say A, or the concept is appropriate, but not the large population study. I don't think that's what you were implying.

DR. GUTTMACHER: Absolutely. I'm not even sure what "the" would describe.

DR. McCABE: Right.

DR. GUTTMACHER: Also, I'm not sure that there will be competing things that come up. But whether they do or not, I'm not going to endorse anything that the committee hasn't even seen, but endorse the concept of such a thing.

DR. McCABE: On the other hand, I think we should hear what are the U.S. activities in this area, and perhaps also have some folks from other countries around the world who may be a little further along than us.

Certainly, the U.K. BioBank has had a number of speed bumps along the way, some of them several stories tall, from what I understand. I think it would be good for us all to hear some of those lessons.

I think having heard David Korn, and I'll now give a personal opinion on this, and seeing what other populations around the world are doing, and knowing that there may be intellectual property that flows from this, this is the business plan of DeCode genetics, that it is important for us to recognize what these issues are, and I think important for us to know what is going on in this country, and to recognize the importance of this in terms of intellectual property.

We, the American people, will own our genes, or the intellectual property flowing from our genes in the future. So I would endorse that we need further discussion of this.

DR. WILLARD: A question I would pose for the committee is whether we should plan such a session if we were interested in one now, or whether we wait until we have some communication from the HHS group so we have something to sort of reflect off of, and look at that potential proposal and concept paper, and then say well, what it may be missing is this. And that is where we need information or where we don't need information. I would throw that open to the committee.

DR. McCABE: Again, I don't think we're in a position to reflect on the science of the proposal. I really think that there are excellent people doing that.

But I think if our charge is genetics, health, and society, we can bring not a broader view, because there is the breadth of expertise within the agencies to deal with this, but I think it would bring a more public discussion of some of these issues related to genetics, health, and society. I think that would be really one of the values in this presentation.

DR. WILLARD: My concern would only be trying to avoid duplication of effort when we have plenty of competing things on our plate for the coming meeting or two.

Alan, is there any written summary of who the actual group is that is exploring the science here? Ed professed great confidence in it, but I don't know who or what "it" is.

DR. GUTTMACHER: I don't know that there is a written list. I can tell you that for instance the working group did involve having folks from both U.K. BioBank and DeCode participate in some of their working group meetings. The working group consisted of both people from within the federal government, and outside experts, some of whom I noticed were suggested for the panels on this sheet. So, you know, it is general expertise.

I don't think personally, I mean, the committee should clearly decide this for itself. The kind of meeting you have on paper here seems like a logical way to inform the committee so that everyone will be approaching it with a fund of knowledge. I suspect a lot of people on the committee already have a fairly good fund of knowledge of this, but others may not.

So to have that first, whether ideally again, we would have something more for you all to react to or whatever at this meeting. But if not, you would still have done the groundwork I think of preparing the committee well to be able to consider it perhaps at the next meeting kind of thing.

DR. TUCKSON: We also need to be, I'm reminded from Sarah, that there is a more formal relationship actually between our committee and your work, and that's Chris Hook who serves on that as our liaison to the effort. So it is no more formal.

So in terms of bringing this to closure, I think what I've got on my notes is that, and let me see if this is what you're proposing, is that we send a letter to the Secretary saying that we have indicated this is one of our priority issues, that we believe that there may be some good possibilities, and there may be benefits that are derived from a well done population-based study that we are urging that the feasibility of such a population study be undertaken urgently, and that we are aware that in fact some efforts may be underway to do that, and we hope that that will be done expeditiously and with attentiveness from his office.

Let me just throw that out as a stocking horse, and you guys beat up on it.

DR. WILLARD: Comments? Debra, and then Ed.

DR. LEONARD: I didn't think we were quite ready to send a letter yet. I thought we were going to have a meeting first to inform the committee and get a better idea of whether we wanted to write a letter or not.

In looking at what is proposed here, it is quite thorough. I would ask that when you have the international cohort individuals come, there may be some information they could provide about cost. Also depending upon how far along these are, what are they learning?

Is the effort scientifically and medically productive, and useful? So some of these are further along. Is it worth the money that they spent doing it? And asking also Ed's question of how are they handling the IP issues when they are making these discoveries?

A lot of the people invited seem to be more academic-based. I don't know if there is a good balance. I know there are a lot of industry efforts in this area.

DR. WILLARD: Just to repeat myself, these are simply what if names. No one has been invited.

DR. LEONARD: Oh, no. I know. But looking at the list, it does seem to be much more academically than industry-oriented. I think there are other industry efforts that might inform the meeting.

DR. WILLARD: I think the question you raised, Debra, whether we should write a very general support the concept letter now, or wait until after we have been better

informed, the reality is if we're going to even have half or a one-day meeting, we are looking at June of '05 before we can actually have such a session.

Is that true, Sarah?

MS. CARR: It sort of depends on what the committee decides regarding pharmacogenomics, the next high priority issue. Then we wanted to conclude with some other possible topics for February. So we'll have to kind of add up.

But I do think that you all have to make some decisions about priority topics for the meeting. The other thing I wanted to say about the letter to the Secretary, it is not the same thing. But the Secretary will know as soon as he receives your road map report that this is a topic of great importance to the committee.

So you can rest assured that he will be aware of that. It doesn't get exactly to the endorsement of the concept, but at least he's aware that we're looking at it. If you decide to do that.

DR. WILLARD: Other comments from the group?

Ed?

DR. McCABE: Yes, I just wanted to lay out. The concern, when I mentioned about competing initiatives, a topic that I am almost as passionate about as genetic discrimination, is children. So one of the issues that I would definitely want to be sure, because I know whenever there are competing initiatives, children usually lose out.

So part of my concern is that we not endorse one initiative knowing of course that that will mean that the kids will not benefit from a single initiative. I just wanted to lay that out, knowing that depending on decisions that are made and my term on this committee, I might not be here to express that passion as loudly as I have the antigenetic discrimination passion.

DR. GUTTMACHER: If I can just join as another member of the American Academy of Pediatrics. The National Children's Study which Ed is referring to, is obviously a very important concept. Again, I should assure the members of the committee that the folks looking at this at the NIH have included people that are intrinsic to the National Children's Study, and we really have been looking about how you might have both a children's study and a study that looked at older individuals as complimentary ones.

I think it underscores the need to, at this time, if the committee does decide to write any kind of letter, to be done that just endorsed the concept in general.

It also would be fair, while I've got the floor, to say if you wanted to say that, you know, any effort to look at this should be cognizant of those efforts done already in other countries, we have tried to take steps to do that. But I think whether we had or not, that would certainly be wise counsel, as Hunt and others have been implying before, and not act as though we are inventing this for the first time.

I think also the other thing that we've done a lot of is looking at how might one interdigitate this with various existing and/or planned cohort studies that are already there, so not to be duplicative.

DR. WILLARD: Emily?

DR. WINN-DEEN: So I just wanted to ask if there were any minutes of your previous meetings that were available that we could get sort of compiled so that we have at least the background information on where you are to date with that program?

DR. WILLARD: There are no publicly available minutes. There are some

notes that the working group members have, and that is all there are.

DR. McCABE: And I think that is one of the advantages of bringing this topic to this committee, which brings it into public discussion.

DR. WILLARD: Brad?

MR. MARGUS: I'd just make a small suggestion that I think the Women's Health Initiative might be a group that you'd want to invite, too. I understand they've collected about 165,000 samples or something. They must have tremendous experience.

DR. TUCKSON: Hunt, can you just clarify that in terms of where you see where we are in terms of our decisionmaking here? Are we sort of saying now that we want to have a meeting? Do we want to start trying to plan for that? Are we waiting? Is it sequential based on other work? Where are we coming to a consensus?

DR. WILLARD: I mean, I'm certainly hearing some enthusiasm for a session. I might suggest that we table the discussion at the moment, go on to pharmacogenomics, which is a little more mature in terms of our thinking and the work of the task force, make a decision on that, and then come back to this if in fact there is room at the February/March issue for at least a half a day to have some discussion.

DR. TUCKSON: All right. We'll put a comma in this, and we're going to just put the pharmacogenomics ? - before we do that, though, let me ask Sarah. I want Sarah to present what is on the board now. I just want to make sure that everybody has in front of them, and this has now already changed based on what happened on the reimbursement one.

You have this little chart at your table. But it is basically just trying to informally and quickly sketch out what is on the plate now. Which things, you know, what the sequence is, which things are in play now that will go away, and by what date. Then wet your appetite for new things. If you can just keep this in the back of your head, and then we'll move to pharmacogenomics in just a second.

Sarah, can you just walk us through this?

MS. CARR: Sure. This is a timeline that on the left side shows all the 12 issues that the committee identified as high priority issues in March of '04. But it also begins with the committee in June of '03. It shows in that month you decided to send a letter on genetic discrimination.

So at each committee, the action or the focus of the meeting is listed there. And then at the March meeting where you established the study priorities, we have shown what else you are doing. The line going to the end is where you have indicated some interest continuing throughout the life of the committee.

We have sort of taken care of genetics, education, and training through the resolution. Although I do think the committee is interested in continuing to be informed about how that is developing. So perhaps I should have continued that arrow beyond.

Then on patents and access, we had decided in March to defer, as Reed said earlier, further work on this until the Academy study was completed. So for the month of October, this year we are showing that you gathered information on that, and then we're waiting for June '05 for the Academy study.

On oversight, the decision made in March, and we had some information gathering in October of '03. We heard from FDA and CMS about the regulation of genetic technologies. We also heard about pharmacogenomics there, the regulatory aspect of that.

The decision was made that this is a high priority issue, but the function we

would undertake in relation to it was to monitor. So the line goes we'll have opportunities throughout the life of the committee to look into what is going on in that issue.

The vision statement has been translated into a report called the road map. We'll be sending that to the Secretary shortly. That will sort of take care of that, I think.

Then we also decided at the March meeting that there were four issues that we thought were high priority that required in-depth study. These four are listed here. For coverage and reimbursement, we have been working hard at that. We gathered information in March, and then (inaudible) the report, and are continuing.

It seems that based on the decision earlier, we will actually probably, I mean, it is possible that we may not actually have a final report until October. It's possible, I think, because it will want to go out for public comment after February, I would think. So that kind of got slightly modified.

And then large population studies in pharmacogenomics we're reflecting here that we are in the planning stages at this meeting. The direct-to-consumer marketing issue is a high priority issue, warranting an in-depth study. We took an initial step of drafting a letter to the Secretary indicating some initial concerns on our part, and requesting that the Secretary take a couple of actions, including charging the relevant agencies to work with FTC, and also to gather data on the public health impact of direct-to-consumer marketing.

Then the three issues at the bottom, access, public understanding, and genetic exceptionalism, were issues that the committee decided sort of were overarching or inherent in all of the other issues. So they are shown here with a dotted line. In all of our other activities, we are trying to highlight these aspects, for example, on coverage and reimbursement, you'll find discussions in the draft, the large issue of access, public understanding, and exceptionalism.

DR. TUCKSON: Great. That's exactly it. So I just wanted you to keep in front of you, because this is an extraordinarily hardworking committee. I just wanted to make sure you don't overload yourselves at any point in time. You see the sequence of things, how they sort of lay out.

With that background, let's turn to the pharmacogenomics, and then we will assess how we will deal with that and/or the population studies one.

Emily?

DR. WINN-DEEN: Well, our task force did meet. I assume that is because we had a staff person who is a great organizer and made sure that we actually had a conference call.

But in the process, we went through a discussion of sort of the first draft of potential things and came up with what you have in your table folders today about potential areas to discuss.

I'm not going to go through them all one by one. I guess in summary what I'd like to say is it seems like there is a lot of meat here, and it is also an area where there is a lot going on that we may either want to just continue to be informed of, or at some point, take some more affirmative action to either support things specifically, or provide some feedback to some of the programs.

What we tried to do in terms of setting out potential topics was to group them into four areas. Setting the stage was basically designed to get everybody on the committee up to sort of the same level of basic understanding.

Translational efforts was basically, again, sort of state of the art. Where are we? What are the issues that we're facing? The ELSI issues again, you know, are there specific ELSI issues that are unique for pharmacogenomics? What might those be?

And then finally when we started listing all the government agencies that we thought HHS had purview over and were involved in this, the list started to get pretty long. We felt from the point of view of the task force at least, that there definitely was some meat here, which because of the involvement of so many HHS agencies, might make this a good area for our committee to spend some time on, and try at least to assure that there is coordination of effort and knowledge of what is going on in the different arenas.

So with that introduction, I'm happy to take any comments and questions. I put Dr. Gutman on notice that we might ask him about some of what is going on at the FDA level, because there are some guidance documents under development and under comment.

I know that there are things going on within NIH. I'm not sure if they are specifically at NHGRI as much as in the Institute of Medicine, or NAS.

DR. GUTTMACHER: The Institute of General Medical Sciences.

DR. WINN-DEEN: General Medicine. But there are definitely things going on within the agencies that I think would be of interest to this committee. The question is just should we let those sort of trickle along or do we want to take some proactive stance on that?

Comments? Ed?

DR. McCABE: Yes. I think there is one topic, and maybe it is buried in some of these others. But as a department chair who was threatened with a pharmacogenomics legal suit a couple of years ago because of a child who had a hearing problem and had received an aminoglycoside antibiotic, I think there is also a medicolegal issue related to these drugs.

When I speak about this, I really argue that the lawyers are going to push this probably more quickly than the clinical side of things.

I don't know if that is something, maybe we don't want to bring that up. But I would think that we have to look at what the medicolegal implications are of ignoring this area.

DR. WINN-DEEN: Right. So that seems to me that that fits under the ELSI issues. But maybe what we didn't cull out was specific, you know, physician or drug company liability if a test was known, available, and not used, and then there was an ADR, what is going on in that front which I think maybe the guys from Pfizer might have something to talk about.

There has certainly been a lot of press recently on several drugs where there may be some genetic component that predisposes people to bad reactions.

Agnes?

MS. MASNY: I think also to dovetail with our discussion this morning about the coverage in reimbursement where we were looking at sort of the overriding principles that might help look at specific genetic tests, and I think the area of pharmacogenomics will probably be the area that is going to advance most rapidly.

So that I think it would be helpful to have some feedback to the committee regarding that, because I think this will be the area where we could then help move the topics of coverage and reimbursement along specifically with the issues of looking at what do we need then to have in place for the government agencies for private insurers to even know when

a pharmacogenomic test will be ready to be used.

DR. WINN-DEEN: Steve, could you comment just for scheduling purposes on when some of this sort of guidance revisions are coming along, and when would be appropriate to maybe hear about those?

DR. GUTMAN: Sure, sure. There actually is a fair amount of activity, both in the Diagnostic Center and in the Drug Center at the FDA. We have published a document on general diagnostic uses related, actually not specific to pharmacogenomics or pharmacogenetics, but to the platforms that support testing in this arena, which would be multiplex products.

We published that about 18 months ago. We have gotten some useful comments, including the thought that a single document is trying to do too much, and that the issues were ? - and we appreciate it. As a result, we are moving forward with a more narrow genetics-oriented document from pharmacogenetics, which we are hoping to publish by the end of the year. Although I hesitate to be certain we'll meet that timeline.

We have within our office a working group. Joe Hackett continues to take the lead. That is very aggressively interacting particularly with industry, but with other academic and government groups to try and educate our core review staff so that we'll be prepared for the diagnostic nuances of this new technology.

In the Center for Drugs, Dr. Lesko is taking the lead. They have published a document encouraging the submission of voluntary data sets. I actually am just absolutely flabbergasted, because I didn't think anybody would submit voluntary data sets. But in fact, that program does appear at least to be generating some light as well as heat.

There are companies that are willing to come forward and share information. There is a working group that is being established to deal with the companies that are brave enough to do that, and to create the appropriate firewalls and controls so that the companies won't be harmed. There are efforts to create an interdisciplinary working group and to create SOPs.

That guidance document establishing the core of that program is also being revised with a target time of being the end of the year. Again, I don't promise it will actually make that deadline, but I expect both will be early next year.

We have recently initiated an effort which is not centrally focused on pharmacogenomics or genetics, although certainly it plays heavily off of them, which is the issue you raised before about theranostics. There has been a long effort between Drugs, Diagnostics, Biologics, and Devices to better coordinate diagnostic and therapeutic products when linked.

It has been a colorful, problematic, and imperfect past on which to build with some interesting successes, and also some interesting failures. There was a workshop actually that was held earlier this year over the summer with PhRMA, with CDER, and with CDRH, Devices and Drugs, all collaboratively involved. There may be many people in the room who were actually present at that meeting.

There was general discussion about the science that underpins how to connect a new diagnostic, or for that matter, an old diagnostic with a new drug or an old drug. And based on a very rich and interesting discussion, there is a joint working group that involves all three human product centers, Biologics, Drugs, and Devices.

For us, it is an aggressive guidance document timeline. It is hoped that we

will complete the internal draft of the document by the end of this year, and have it vetted in time for the third pharmacogenomics workshop, which Drugs will be sponsoring in April of next year.

Although I suspect that workshop will have a wide variety of topics on its agenda, certainly front and center, and from my perspective, the most important, will be vetting of the joint diagnostic therapeutic guidance document.

And then most recently, the Agency has promoted as part of its perhaps in the wake of Dr. McClellan's influence, its interest in making sure that it is a partner rather than an impediment in translational research. They have introduced a program called the Critical Path Program which is to actually in a more proactive way seek out ways to bring products to market more quickly, and pharmacogenomics has been targeted as a pilot, or as an opportunity within the product lines that we regulate to be exploited in a positive manner.

DR. WINN-DEEN: Ed?

DR. McCABE: Yes, I think about this. There is some overlap between the two topics we're discussing, large population studies and pharmacogenomics. Because to get at the genetic basis for rare side effects will take large populations.

I don't know, it sounds like these are somewhat competing, and I don't know if there is a staging. I don't have an answer to this. But perhaps there could be some discussion of staging if it is better for one to go before the other.

DR. WINN-DEEN: Well, it certainly sounds like there is a lot of things going on actively that might be ready to make some report on at the whatever it is, end of February or early March meeting. At least from the regulatory side of things.

Alan, do you know in your large population studies, was there a pharmaceutical component to that? Or was it more trying to understand the genetic components of common complex disease?

DR. GUTTMACHER: Well, the folks are clearly understanding genetic and environmental factors and common disorders. Very much the thought is if one is going to do a large study, it would be foolish not to take advantage of that in this day and age, to look at pharmacogenomics issues as well. Absolutely.

DR. WINN-DEEN: Steve, I don't know, you're probably not the right FDA person to ask this, but I'll ask you anyway. Do you know for the two big drugs that have been taken off the market recently, if there is any feeling that they have some possibility for rescue based on any kind of test segmentation?

DR. GUTMAN: Yes, I actually don't know.

DR. WINN-DEEN: I think this is an area of fairly substantial concern about trying to understand what health risks we are putting people to, and whether there are simple tests that could identify who those folks at highest risk are.

MR. MARGUS: Emily, one thing that came up I guess last March maybe when we were still discussing whether to make this a priority, or there was a discussion about pharmacogenomics, and we did have some visitors, right?

With regards to CMS, besides the concern that tests would be covered, the other issue is that if hundreds of millions of dollars are being spent reimbursing people for drugs of which 40 percent of the people who have taken the drugs aren't actually responding, there is a real economic value to pharmacogenomics that CMS and certainly the private carriers should be very interested in. As a different aspect of how CMS might be interested or how we

might encourage CMS besides just covering the test.

DR. WINN-DEEN: Any other comments?

(No response.)

DR. WINN-DEEN: I know we're short on time, so I'm going to yield the floor.

DR. TUCKSON: Thank you for leading that, Emily.

Let's just remind ourselves that if you go back to your little grid chart here, as Sarah took us through it, first, the genetic discrimination issue, we've got a lot of work that we're going to be doing between now and the next meeting, and probably we'll have something on the agenda around genetic discrimination. That is going to occupy some considerable attention. So that is an active ball in play.

The genetic education and training is probably not a ball in play, but certainly the next ball in play that is serious is the coverage and reimbursement. That is going to take a lot of our time at the next meeting, and we're going to have to nail down some considerable work there. So those two are big.

The other thing that is big, and let me just make sure I've got it in my list. I'm keeping track. Let's see. Those are the big ones. We're not going to wind up doing much on the intellectual property one until after the other committee meets.

So we really do have the opportunity I guess to really start to think about do we want to grab onto for the next meeting, both or one of the large population studies of the pharmacogenomics, and mature those. Then we have these other potential topics as well.

Thank you, Sarah. Why don't you take us through those, Sarah?

MS. CARR: Well, the first one is reflective of an interest expressed by the committee in June, and it sort of came up I think in the context of the discussion of education and training resolution. It was some rather significant interest expressed in what is going on in health information technology.

So we just wanted to make sure to just put it on the table here for the committee to decide whether you would like to be briefed in a fuller way about what is going on.

DR. TUCKSON: Let me, on that one, just say that given the discussion we had at this meeting regarding the family history, I think that to leverage that discussion, we may want to have a brief update on this. Clearly the implications for the electronic medical record and all the technology stuff that is going forward.

I will tell you from some other hats that I wear, that whole area is moving so rapidly. The idea of creating standards for the electronic medical record is moving from yappa, yappa, yappa, to implementation at rapid rates. So given that we spent as much time as we did today at this meeting on the family history stuff, we may want to well start to connect those in and leverage our time.

DR. WINN-DEEN: I just wanted to say that also has a lot of relevance to the pharmacogenomics. Again, this is information that you would be tested for once, and then that information would be good for the rest of your lifetime.

DR. TUCKSON: So we may want to well try to squeeze in at least a presentation on this.

MS. CARR: And then there is a third aspect to it, too, which Ed McCabe might want to speak to. That is that the way in which electronic health records might help

enhance privacy.

Ed, I think you mentioned that you had had a conversation with Mark Rothstein about the growing realization that rather than diminishing privacy, it might actually enhance it. So that might be a way, an aspect of it.

But the second issue is the presentation from the HRSA Committee on Newborn Screening about the recommendations they are making to the Secretary. Chris Hook, for one, was very interested in having such a presentation.

DR. TUCKSON: And given that we are, again, officially liaison to that, and given that that has come up pretty directly today, I think this committee really does need to hear at least a brief update on that.

DR. FEETHAM: And the word I had in talking to Dr. Puryear that that would be timely, and that report would be ready.

MS. CARR: Thank you for checking on that. Then the last two are presentations that we could request of CDC on rare disease testing. There was a conference, a public/private-sponsored conference, that came up with a lot of recommendations about how to enhance access and quality genetic testing for rare diseases. That group is interested in presenting their recommendations to this committee and getting some sense of your perspectives about them.

And then CDC could provide a very quick presentation on some of the work that they are doing to enhance quality assurance and quality control through the laboratory services program.

Is Joe Boone here? In case he wants to say anything further about those two topics. They were suggested by Joe.

DR. WINN-DEEN: So I have been involved in those things. I think they would be quite germane, to at least just get an update on what they are doing to try and fill what is a really big void right now for genetic testing.

DR. TUCKSON: Right. So let's do this, then. We are going to finish on time, which is terrific. So let's slow down a minute and look at all the stuff we've got in front of us.

Let me remind us as we consider it, we've got four things that are there. We've got pharmacogenomics, and we've got population studies, so keep those there. That's six. Now, what we did this meeting, my quick list here. I can't believe how much we actually did, it's amazing.

First, the family history project. We are going to get back from the family history project people what all the public education materials that are going out that are relevant to genetics education, and how they intend to connect the family history project to the electronic medical record, and particularly all this HIT infrastructure.

We are sending a letter to the Secretary endorsing the importance of family history as a tool. We are saying to him that we are encouraging to see HHS agencies playing nice together, and that we are encouraging those who are not playing nice, or who aren't involved, to get involved. So that is one set of things which we have done, straight forward.

Number two is genetic discrimination. We have agreed that we are going to compile the testimony from today, including the names of the people that presented, and their congressional districts, and a slew of background materials, apropos McCabe. We are going to put all that together, and then ask to meet with the Secretary to discuss this stuff, as well as to

have him convene at the same time the leaders of Justice, Labor, and Commerce, so that we might try to resolve any differences that may exist within the agencies.

EEOC, by the way, is going to analyze gaps in current state and federal legislation. We are asking the Secretary to send this compilation testimony to Congress, focusing on the Speaker, the Majority Leader, and Congressman Barton. We are going to ask the Gene Coalition to send us their membership and analysis, and what it would take from their point of view to solve the problem.

We are asking AHIP for their analysis of what is wrong with the Senate legislation and why they can't get on board, and what is necessary to solve the problem. We are sending a letter to the Coalition for Genetic Fairness requesting clarification on whether they are opposing the use of information for care coordination, or whether they have some other issue that prevents them from getting to agreeing on a consensus.

We are considering bringing either congressional people or senior staff here for the next meeting to see whether or not that will help to try to understand better what it is going to take to get to success. We are considering holding a roundtable discussion to get consensus of key parties at the next meeting, i.e. the Chamber-type people, and the coalition people. So we are trying to think maybe that may be necessary to do at the next meeting.

We need a conference call to discuss the next steps so that we'll know basically how to proceed in this regard. It it something we'll have to do between meetings offline.

The third area was coverage and reimbursement. We are making genetic counseling a priority. We need to deal with the licensure issue and scope of work. We agreed to do a literature review and analysis before the next meeting. Cindy is taking charge of that. Muin is supposed to help. The National Society and Board of Genetic Counseling are providing their input. As far as the overall reimbursement report itself, we've got a significant more amount of work to do, and they will be sending next steps back to us.

On the intellectual property rights issue, we basically have said that we're going to wait for the final report from the NAS committee, and based on that, how do we deal with the issue of how that affects the protection of the health of the public.

Large population studies we are going to discuss. Pharmacogenomics we are going to discuss.

Man, we did a lot of work. Now, what did I miss, by the way, in terms of what we did?

DR. McCABE: The only thing you forgot on the genetic discrimination is to invite my friends at the Chamber to come and appear before us.

DR. TUCKSON: Done. We'll get that. And then not your mouthpiece, if I recall. So the Chamber gets invited, terrific.

Now, somebody's pet peeve or issue I didn't say. So other than that, what did I miss in terms of what we did or committed to this meeting?

DR. McCABE: I think it was very thorough. I would encourage you, I think it's time for consolidation of what all you have accomplished. Rather than taking on another major topic, pharmacogenomics, or large population studies, I would encourage you to use the next meeting to consolidate and move forward.

I think there is an awful lot on the plate of this committee. You know, I would be concerned that taking on another major topic would be too ambitious.

DR. FEETHAM: Also, as part of the discussion from the reimbursement study, we had talked about the fact, or the reimbursement report, we had talked about the fact of hearing back from the HRSA study, that that may inform that discussion.

DR. TUCKSON: I'm sorry? Say it one more time.

DR. FEETHAM: As part of the reimbursement report, we had talked about the fact of hearing the latest report back from the HRSA genetic services study to see if that would inform that work.

DR. TUCKSON: Good. Thank you. So what has happened is that Ed in his chairmanship-like way has moved us from any errors of omission from my summary of what we decided to analyzing that and saying his conclusion is that we've got a lot on the plate, given that we had these other two issues here, that we did agree that we wanted to get that brief update on the newborn screening, which I think is important, and we wanted to get the briefing on the informatics initiative. So those were two things.

He is saying, I think, then that he is not urging that we bite off the pharmacogenomics or the population study, other than, well, he is saying right now -- let me not reinterpret what he said -- that we bite off those two quite yet. Let me just see if there are other opinions on that.

DR. WILLARD: Just a point of information. I mean, there are two presentations there on newborn screening, and on testing for rare genetic diseases, which are clearly for the information of this committee, but I'm trying to figure out whether those are in our critical path to get to some action item.

Whereas pharmacogenomics and large population studies clearly are priority issues that we want to at some point weigh in on. If we have to make tough choices, it may to me hinge on whether we have future action items where we absolutely need certain information.

DR. TUCKSON: Let me try it this way, and Hunt, you're terrific.

What I'm thinking is that for the brief informational item ones that just take a few moments to give us a little update, they don't imply another set of work, whereas the pharmacogenomics and the large population studies are saying that there will be subcommittees that will continue and go forward.

Now, the sequential nature of this may be, Hunt, and maybe that's what you're getting at, is that we can at least have some more maturing of those projects, and then put them into the queue, because they'll start to be massaged offline.

DR. WILLARD: Well, that was my sense. If we could devote a certain couple of hours to pharmacogenomics or large population, that would allow the task force to actually jump into action between meetings. Whereas if we skip a meeting, the task force can't do anything anyway. Then we've basically lost four to six months.

DR. TUCKSON: Other points of view?

DR. McCABE: And if you wanted to move one of the topics forward, I think perhaps a little more in-depth briefing on what is going on in HHS might be a way of occupying an hour, but more to keep the momentum going in these directions. I would leave that up to the task force.

DR. TUCKSON: So that would say, and Ed, in that comment, are you saying that both of those have activities going to HHS, or did that sort of imply that you want to sort of get something from HHS on the large population one?

DR. McCABE: Well, I know that there is stuff going ? - I mean, there are plans afoot in HHS for the large population studies. I don't know if there is anything quite so concrete, or something that is easy to get one's arms around in the pharmacogenomics. Perhaps we could be informed.

DR. GUTTMACHER: I would think it might in some ways be close. More equal than you're implying. In fact, I think there are not plans afoot, they are simply exploration afoot. It hasn't really gotten to the point of anything close to plans, I would say.

DR. TUCKSON: Well, let me see if I can take what the guidance is from you and give you one more chance to react to this. I think that what I like from Hunt is that we don't want to lose the momentum of those committees. We don't have to dedicate ourselves to solving both of those issues tomorrow.

What we need to do is to have the committees create at least some time for there to be more discussion and discovery at the next meeting, which then will allow this to go on. By that point, by the end of the next meeting, we'll be ready to start to tackle some of these two. Basically you want to keep sort of both of them alive and moving. It is up to Sarah and I to try to figure out on the agenda how much time we can give to all of this.

If that is your recommendation, let me see how your colleagues feel about that.

DR. WINN-DEEN: Well, just from the pharmacogenomics thing, I think we probably have two or three meetings worth of stuff that we could talk about. So I wouldn't have any problem if we split that up and did a couple of hours here, a couple of hours there, just sort of the latest, hottest things going on.

DR. TUCKSON: Well, I don't see any strong objections, so I think Sarah and I will work hard to get all of these on the agenda and give them the appropriate amount of time. But I think with Ed, I think we are pretty straightforward that we're going to knock out the ones that we've got pretty well mature, get those things really resolved well.

In the interim between the meetings, there are two other things I want to bring up quickly. One is we will send you in the mail a copy of this from the General Services Administration. I really was curious about how you evaluate the quality of a committee like ours. I was very surprised to find quite a very thoughtful analysis that is used to determine whether or not we are worth diddley-squat.

So I want you to see that and keep it in front of you. It does sort of give you a little reality check, as it were, about what we're doing. By the way, I think given Ed's leadership in the past, we'll comport ourselves fairly well in that analysis. But I want you to keep that in mind.

Secondly, I wonder whether I could have your permission to draft a letter that I would send on your behalf, of course you'll write it, that would urge the Secretary to create, and this is not the right word, but sort of a czar coordinative person. Some identified single entity at HHS for these genetics kinds of things. We keep bouncing all over the place with this thing.

Quite frankly, while I am comfortable that before I would take this position that I did talk to the Secretary's office, as I mentioned in my opening remarks yesterday, that this was important to the Secretary's Office, I think that I would feel even better if there was a person that was sort of designated to receive our work, to know that they were on point, and to serve as an official coordinator across the agencies for the things that we're talking about.

I just feel like I would like to make that sort of a little more explicit and more direct. But I open the floor for your guidances as to whether this is worthwhile.

DR. McCABE: So moved?

DR. TUCKSON: Argument? Other points of view?

DR. WILLARD: Would your intent be to circulate that and send it quickly? Or circulate it and have a draft for final decision at the February/March meeting?

DR. TUCKSON: First, great question. Again, just as a point of departure, since there are several things that we're sending to the Secretary as a result of this meeting, I would probably put it in that letter as part of the overall letter and say you know what man, you've got a committee that is working its tail off on issues that are important to the American people. They range in quite a variety of areas. As such, I think we'd like to know that there is somebody who is paying attention to these issues. So it would be packaged into our report to him on this meeting.

All right. I'm not hearing any violent opposition to that, so you'll be seeing that as well. Who on this committee has any further thoughts? Anything that did not get discussed? Miscellaneous? Yes?

MS. HARRISON: I get increasingly concerned that the public doesn't necessarily know everything that we're doing. So in light of that, in a previous discussion that was some talk about putting that vision statement priorities report into at least the scientific literature. If not, even trying to coach it into the lay person's literature, just so again, we could increase awareness about our activities and maybe even get more public comment than we do already from folks that don't traditionally read the Federal Register.

DR. TUCKSON: Barbara, I think you're really on to something there.

MS. HARRISON: And then just one second thing is the testimony yesterday was just so powerful, and again, in this atmosphere of people saying that genetic discrimination doesn't exist and there is nothing in the literature to support that, I think the information that we got yesterday, I'd just throw it out there that possibly we could coach that into something again, that could go into the scientific literature so that people can reference it in the future.

DR. TUCKSON: Boy, those are terrific comments. I was trying in a way, Barbara, also not as eloquently as you, when we had those folks who were testifying, I would sort of say, is there a way in which you can take back to your newsletters and so forth and so on. I think what we ought to try to do is to try to formalize that a little bit, and the major advocacy organizations for the genetic community is to request that synopsis versions of what we're doing, or whatever, are made available to their membership.

I think we should actively, if we have not, Sarah, just formally reach out and say we really would like you to do that. I saw some other hands. Those were terrific.

Emily?

DR. WINN-DEEN: I just wanted to ask if we were going to comment or vote on this draft direct-to-consumer letter. Are we going to give you our blessing to send that? Because it is in the table of packets, but we didn't really talk about it.

MS. CARR: Well, we thought we had your blessing to send it. We had sent it out for comments electronically. As Reed said in the very beginning, we are more than happy to get more comments about it.

DR. WINN-DEEN: I'm just trying to drive it to conclusion. Are we done

with it? Are we ready to send it?

DR. TUCKSON: The answer is that you have until the end of this meeting or by the time Sarah gets back to the office tomorrow to send something in. But absent that, it is going out.

But the answer is that you do have time. So if you forgot to read it, didn't look at it, don't remember it, yes, you still have a moment to comment. Other points? These are terrific.

DR. LEONARD: Mine was the same as Barbara's. We are sending the road map to the Secretary, but we had also discussed publishing it. I think that is a very, very important step.

DR. TUCKSON: All right. We'll work on that. Well, listen, let me thank all the members of the audience who have ? - I mean, there are some of you who have been here the entire two days. You are extraordinarily resilient, and we thank you for being part of our little community.

To Sarah and the team, good Lord, my goodness. You all worked something fierce. It is very much appreciated. The support we get is embarrassing.

So thank you very much to everyone. Good meeting.

(Whereupon, at 3:09 p.m., the meeting was adjourned.)