Discussion and Plans for Next Issues

DR. McCABE: We're going to go ahead and get started. I apologize to everyone for rushing everybody's lunch, and especially I apologize to Joan.

The first topic is going to be the resolution on genetics education and training of health professionals. It's been revised since yesterday, and it's sort of been undergoing continuing revision, and I'll let Joan take the lead.

Again, I'm sorry, Joan.

DR. REEDE: No problem. While we're waiting for it to go on the board, I wanted to make a few comments first. Thank you to the staff for helping us get these revisions in, and to members of the task force, who met this morning before our meeting started to go over some of those revisions.

Secondly, I wanted to make a statement in general about some of my philosophy on wordsmithing and what we're trying to accomplish. One of the things I think that can sometimes be difficult with these kinds of documents is when you try to be all-encompassing and to do everything with one document. So after a while, you actually start to lose what was the purpose of the document. So I think we have tried to capture the conversation from yesterday, the edits from yesterday, and at the same time tried to make this simple and direct, with an understanding that we can come back and this does not have to be the final time that this committee speaks on education and training, and that if there are areas that the committee would like us to explore further or go into more depth in the future, that can be done, as opposed to trying to do everything now.

Now, I'm going to do what I did yesterday, and I had not known that I was going to have to develop skills of looking behind me and in front of me and twisting and speaking at the same time when I came on this committee, so let's see if I can figure out how to do this.

I'm going to go down them, read them as the are. If there are comments, I'm going to try to bring them up as we go forward. If they are small changes, if you could hold on to them. I'm looking more for general concepts that we're off on.

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

"Whereas advances in genomics will lead to a more precise understanding of disease processes and will provide better guidance on the application of therapeutic and preventive strategies that will make significant improvements in health status and outcomes;

"Whereas insufficient education and training in genetics and genomics has led and may continue to lead to inaccurate or delayed disease diagnoses, misguided disease management family planning, increased health disparities, and excessive costs;

"Whereas appropriate and adequate training and education in genomics is crucial for all health care and public health professionals to assure appropriate, effective, and efficient integration of genomic concepts and genetic technologies and services throughout the entire health system;

"Whereas appropriate education in genomics is crucial for the general public to take advantage of the benefits of genetics and genomic advances;

"Whereas education of health care and public health professionals and the public is necessary to assure equitable access to genetic and genomic technologies;

"Whereas education of health care and public health professionals and the public is a necessary component of the application of evidence-based medicine related to genetics and genomics;

"Whereas through a survey of federal agencies on their role and activities in genetics and genomics education, training, and health workforce analysis, it was found that federal efforts are focused on translation and appropriate integration of new genetics and genomics technologies into health

care and public health;

"Whereas a solicitation of information from educational and professional organizations identified the following urgent needs in genetics and genomics education and training:

"Inventoried, catalogued, widely relevant clinical and public health applications stemming from advances in genomics;

"Educational models that use such applications to clarify how genetics and genomics, through the use of family history tools, information technologies, and Web-based practice tools, among others, should be integrated into practice;

"Incorporation of genetic and genomic competencies into national accreditation and re-accreditation standards;

"A broadening of the focus of genetics education and training to incorporate both genetics and genomics;

"Assuring the diversity of the health care and public health workforce and the cultural competence of its members;

"Increasing the presence of faculty appropriately trained in genetics and genomics;

"Training programs that address the interface of an interaction between genomics, their ethical, legal and social implications, and public policy.

"As such and in light of the importance of ensuring that the benefits of the genetics/genomics revolution are accessible to all Americans, SACGHS urges the Secretary to take the following steps to ensure that genetics education training of all health care and public health professionals is adequate:

"Promote and actively incorporate into departmental policies and programs the philosophy that genetic information, which includes family history information, should not be treated as exceptional but rather as part of the spectrum of health information and viewed as an integral part of the practice of all health professionals;

"Incorporate genetics and genomics, including family history tools and point-of-care educational support, into relevant initiatives of the Department of Health and Human Services, including the Secretary's Health Information Technology Initiative, and engage in the dissemination of this knowledge to health care and public health professionals;

"Promote and support initiatives that address the integration of genomics into the education and training of all health professionals.

"In order to facilitate the integration of genomics into health care and public health now, direct HHS agencies to work collaboratively with the state, federal, and private organizations, such as NCHPEG, to support the development, cataloguing, and dissemination of case studies and practice models that demonstrate the current relevance and applicability of genomics to health care and public health;

"Provide adequate program and technical support to federal programs that provide for faculty training in the implementation of clinical application-based genomics education models, particularly models using clinically relevant examples and that incorporate the ethical, legal, and social implications of genetics and genomics;

"Promote communication among all health professionals to enhance the accessibility and widespread dissemination of genomics educational models and applications, and raise awareness among all health professionals, faculty, and professional educational organizations of these resources;

"Work with ASTHO and other relevant organizations to address issues associated with incorporating knowledge of human genetics and genomics into accreditation, licensure, and certification;

"Continue to encourage support and facilitate programs that address the need for

workforce diversity and cultural competency of health professionals, including sensitivity to the disability community;

"Provide adequate support for efforts that will incorporate a genetics/genomics focus into pipeline programs supported by HHS;

"Promote culturally appropriate and sensitive public education that provides the knowledge and skills that consumers require to participate effectively with health professionals in decisions that increasingly are informed by genetic perspectives."

Comments? Questions?

Emily?

DR. WINN-DEEN: So just a minor point. I think NCHPEG and ASTHO should be -- you should say what they are, because in alphabet soup-land, not everybody knows.

DR. REEDE: Okay.

DR. WILLARD: On that same point, I guess I would question why NCHPEG was being singled out. I mean, I recognize that that's something they are doing, but it's not like they're the only organization that has been charged with doing it through some official channels, as opposed to ASTHO, which actually is charged with dealing with some of those issues.

DR. REEDE: I think the conversation yesterday and the general consensus was that NCHPEG should be listed specifically as an example of an organization that is doing this. So that was a general consensus of yesterday's discussion.

DR. LEONARD: In looking at basically our recommendations, many of them say sort of in vague terms what should be done, but I can imagine Secretary Thompson, having presented recommendations to him at one point from laboratory or pathology organizations about genetics and genomics and molecular diagnostic testing, his question was always, with every recommendation, how do you propose that I do that? There are many things on this list that we're not making a specific recommendation. It's relatively vague and philosophical rather than an implementable recommendation.

So, Ed, maybe you've done this a lot more than the rest of us and you could comment on the need to be directly implementable, as opposed to philosophical.

DR. McCABE: I think that there's a role for both, that sometimes if we have mechanisms that we wish to use to recommend for implementation, we should give them, but sometimes it's redirecting issues philosophically as well. So as direct as we can be, we should be.

DR. REEDE: Paul?

MR. MILLER: The paragraph that's up on the screen, to promote and actively incorporate genetic information, should not be treated as exceptional but rather part of the general spectrum of health information and so on, that might flag. That might have an impact if that's the sense of the committee. That might have an impact on the nondiscrimination legislation in the sense that one of the arguments against genetic nondiscrimination legislation is that genetic information is not exceptional, it's just regular old health information. So why should we treat for discrimination purposes the use in privacy and so on of genetic information any differently from anything else?

That is a very strong undercurrent in that debate around the nondiscrimination language, and there are a couple of sort of terms of art or buzzwords in there that I would be concerned if all of a sudden somebody grabs onto that language and says, well, here's the Secretary's Advisory Committee saying that really genetic information should not be treated as exceptional, and therefore undermines this committee's other sentiment around nondiscrimination legislation.

DR. REEDE: Right. I think that part of our discussion in the past had been to look at exceptional based on whatever topic we were looking at in terms of how it would be incorporated. So one suggestion might be that it not be treated as exceptional with regard to education, because I think we're speaking specifically that with regard to education it should not be treated as exceptional but rather as

integral. With that change, would that address the issues that you're raising?

MR. MILLER: Yes, I think that would make it much more clear.

DR. GUTTMACHER: I wonder, though, whether -- Joan? Alan, over here. I wonder, though, is "exceptional" really the right word for this context? I think perhaps it isn't. It's not exceptional versus integral I think in this context. It's the idea that we want it not as freestanding, not as just genetics someplace but that it needs to be integrated.

So I wonder whether we could just get rid of the not treated as exceptional and just say should be treated as part of the spectrum of health information and viewed as an integral part of the practice of all health professionals, something like that.

DR. FEETHAM: That was going to be my comment also. I agree with that.

MR. MILLER: The word "exceptional" is really a buzzword.

DR. FEETHAM: It's integral, that you really want it to be part of the practice.

DR. FELIX-AARON: Yes, I support that.

DR. REEDE: So it sounds like a consensus that "exceptional" gets taken out and what we're really trying to reflect is that it should be integral and not as a stand-alone, separate, optional piece.

Other comments, suggestions, questions, changes?

DR. WILLARD: Just in general, whenever we finally get done with this, there's a lot of inconsistencies, grammatical inconsistencies. The HHS is referred to about four different ways and no obvious rationale for doing it. That's just a staff issue at the very end of the day.

DR. REEDE: If we can leave it to staff to make sure that we're grammatically correct and consistent, and our acronyms are defined, et cetera.

DR. FELIX-AARON: A question, Joan. I don't have the document in front of me, but as you were reading it, I thought that there were many recommendations that we had, and I was just wondering how many recommendations do we have, and do we want to be parsimonious? Again, trying to balance that with the need to have the things that we think are valuable represented in the recommendations.

DR. REEDE: There ended up being 10 recommendations in the end, and that partly came from yesterday's where there were some recommendations that were split from one into two.

DR. FELIX-AARON: I mean, I don't have a clear recommendation, just to share the sense with the group.

DR. REEDE: I think one of our concerns was making sure that it didn't become just a long laundry list that people would not pay attention to but rather ignore. I don't think we ventured into that laundry list territory, but I do think it's something to pay attention to.

DR. LEONARD: Has anybody looked at these and prioritized them so that the ones with greatest significance or impact would be at the top? I don't know that there's necessarily a rule that the ones at the top are paid more attention to, but they might be.

DR. REEDE: We have not tried to prioritize them, but the first step that we took this morning was actually just trying to put them together. So if there were two that related to culture, they actually flowed one behind the other. But we did not try to prioritize them.

I think the consensus from yesterday was that we should start out with the general one that refers to incorporation of these concepts across the various agencies, and that would be the strongest, and then we went from there. Is it the wish of the committee that we try to prioritize these?

MR. MILLER: If I can just make a statement, I'm sort of getting dizzy watching the document jump back and forth. I think I'm on the whirlybird at Disneyland.

What would be helpful for me, since nobody has the printed document in front of them, although I hope nobody has it because then I would feel very lonely not having it, to go through paragraph by paragraph and really tick it off, because it's really hard after the initial read-through and

having it jump around to really sort of sign off on the document or understand the document.

DR. REEDE: I agree. I think we can definitely do that. I think we did a part of this yesterday. My only caution is that we don't extend this into a three-hour discussion of wordsmithing for each piece and not get through it. But what I'm going to ask is the same thing I did yesterday. Sarah, if you could help from there, because this is very awkward for me to try to read around this.

MS. CARR: Do you want me to read it, or do we want to take a pause and get a hard copy and maybe go into the coverage and reimbursement and come back with the hard copy in front of you? We can do that.

DR. REEDE: The hard copy would be easier.

MS. CARR: Do you want to do that, then? Okay.

DR. McCABE: Cindy, are you ready to lead the discussion on coverage and reimbursement?

MS. BERRY: I guess. It's sort of hard to figure out where we left off yesterday. I think we do need to focus, kind of really home in on our recommendations.

MS. SARATA: I have an outline developed for you to work off of.

MS. BERRY: Okay, we're going to get help. I was going to suggest putting the topics up on the screen, but I don't want to interfere with the work that's going on right now. I think it will take us a few minutes.

DR. McCABE: While we're waiting for this to come up, we had a few changes, but the primary changes were in the recommendations and trying to organize the recommendations because they were too numerous. Is that correct? So we got them down to either four or five, depending on whether we considered the broad areas, which were two under the broad areas, and whether we considered that one or two.

The other thing I would remind everyone is that we decided yesterday we would probably not finalize this at this meeting, but we would come up with a second draft of this from the meeting.

MS. BERRY: I think we're ready to go. Staff once again has come through, as they always do. They have put together an outline that attempts to reflect some of the discussion that we had yesterday, because we were struggling with the myriad of topics that we could address in recommendation form. In an attempt to organize that, staff have come up with this outline.

I actually would put something before coverage, and we did talk yesterday about defining what we're talking about when we discuss the term "genetic technologies," whether it's genetic tests, genetic services. What is it precisely we are trying to get covered and reimbursed properly? We need to clarify that a little bit better than we did initially in the first draft of the report.

To the extent that folks have any specific input on that, we should nail that down, I think, because that's really a threshold question. We're not saying cover everything and reimburse everyone no matter who is doing it, no matter what they're doing. We are trying to be focused here. So that will be an important up-front discussion in the report that will be fleshed out a little bit more than it currently is.

I don't know if anybody has any comments on that particular point.

Debra?

DR. LEONARD: But that would be part of the body of the document and not part of the recommendations.

MS. BERRY: That's right. The recommendations, though, will relate to that, because we'll be clear all along that what we're recommending goes back to our initial definition of those technologies.

Hunt?

DR. WILLARD: Again, I'm thinking of the consumer who is reading any of this. It's the juxtaposition of the terms "genetic services" and "genetic technologies" which -- and I believe we mean different things. As defined in the document, "genetic services" applies only to those who are board certified medical geneticists, counselors, or primary care physicians. It therefore excludes any other specialists who might do lower-case g genetics or genomics work, and that's fine. We can define that any way the committee wishes.

But then the parallel term, "genetics technologies," on the other hand, takes on a much more all-encompassing flavor to it and covers everything under the sun, not what would be traditionally or classically considered "a genetic test," circa 10 years ago, but everything that might come forward for the next 10 or 20 years based on the Human Genome Project. So I'm a little concerned that those two terms invite one to treat them as parallels when, in fact, they're very, very different.

MS. BERRY: Does anyone have any other thoughts about that? Because that is really an important up-front matter that we have to tackle before we can really be precise enough to be useful in our recommendations.

Debra?

DR. LEONARD: I'm not even sure we should be using the word "technologies." Maybe "test" is a better word to use, because I see technology as methods rather than a clinical service that's being provided.

DR. WILLARD: It comes from the charter, however, which uses the term "technologies."

MS. BERRY: What about in the beginning, though? We can define our scope how we wish in the report, where we can say in this case or for purposes of this report, we are referring to just this one aspect of genetic technologies, genetic tests. We can narrow it for purposes of the report without interfering with our charter and our other goals and duties.

DR. LEONARD: I mean, as "genetic technologies" is defined, it's correct. They're technologies, but those technologies are used for tests or testing for clinical purposes, and it's really the tests, using a variety of genetic technologies, that may change over time that we're concerned about. The laboratory tests, and then the other medical services surrounding patient counseling and treatment and everything also is what I would put into services.

So maybe what we need to define are genetic technologies, genetic tests, and genetic services.

MS. BERRY: Anyone else have any thoughts?

Martha?

DR. TURNER: Yes. Just a question we were asking over here is that the written comments that we got from people that are in our folders, I wondered if those had been received in time to incorporate them or to consider the suggestions in these documents for that draft.

MS. SARATA: No, they hadn't.

MS. BERRY: There's mouthing going on.

DR. TURNER: The other question is do we need to do that in this group now, or will that happen later?

MS. GOODWIN: Most of the public comments that we received were received after the briefing books but before this meeting, so most of the public comments are in your table folders and they've been reviewed to the extent that the committee members have been able to review them during this meeting.

MS. BERRY: But we will need to consider them and, to the extent possible, incorporate them in the next iteration of this report, of the draft. But they haven't been incorporated in this particular version that people have looked at.

Well, any other suggestions on the threshold issue of how we want to define what we are proposing to cover and reimburse, or should we just have a go at it with another draft and reflect some of the comments that we've received from the public, and then send a second draft out? I don't mean to cut off debate either, because people may have additional thoughts as you go back to your offices, and you should feel free to email some of those suggestions because I do think we need to nail that down very well. I don't want to gloss over that. It's pretty critical to what we're doing.

Kay?

DR. FELIX-AARON: Just a clarifying question. Are we going to go through the different -- number 1, number 2?

MS. BERRY: I just put a 1a before the coverage that's not reflected in the outline.

DR. FELIX-AARON: Okay. Thanks.

MS. BERRY: Emily?

DR. WINN-DEEN: I just wanted to agree with Debra, that I think in the context that most people in our part of the world, in the diagnostic side of the world use it, we should refer to this as coverage of genetic tests and not of genetic technologies, because the technology is just a means to the end, but what you're developing evidence for is that the genetic test has a clinical utility in medicine, and what you want reimbursement for is when a physician orders a genetic test, that that is reimbursed. It's sort of technology independent, with the exception that the CPT codes code reimbursement by the actual steps that are performed; as they exist today, code for the specific test steps that are required to be performed.

MS. GOODWIN: Are you suggesting, then, changing the title of the report to "Coverage and Reimbursement of Genetic Tests and Services"?

DR. WINN-DEEN: So where you say "genetic technologies and services," I think it should say "genetic testing and services."

MS. GOODWIN: But you're still including the services part of it?

DR. WINN-DEEN: Yes, absolutely.

MS. BERRY: Any other comments on this?

Debra?

DR. LEONARD: But that services is to be distinguished from laboratory-developed tests, which in our previous discussion were referred to as services. That's not the services that we're talking about here but more like medical genetics or genetic counseling, treatment follow-ups, interpretation of the test results, those types of services.

MS. BERRY: We should clarify that in some sort of definition section to really nail that down.

Okay, moving to the first coverage section, we had a lot of discussion yesterday about the evidence base for -- really the lack of an evidence base hampering coverage decisions and determinations. So there was a discussion about doing a technology assessment or some sort of study as to really what is out there. What evidence do we have that supports coverage of certain tests or services? We did not go into too many specifics. We talked about AHRQ, we talked about other entities. Some have proposed the National Academy of Sciences/Institute of Medicine as a possibility. There may be others.

Do folks on the committee have a preference or an idea for who should actually conduct this? Do we want to recommend a specific entity to the Secretary in our report for recommendations, or do we want to leave that vague? If we want to have a specific recommendation, what is the preference?

Ed?

DR. McCABE: Recognizing that I am a member of the Institute of Medicine,

however I would press for this being a study commissioned by the Institute of Medicine. I think that's an independent group and it would carry a lot of weight with the Secretary, I would hope, and I think it would, by making it independent of the agencies of HHS, that hopefully it would be recognized as credible by the Secretary. So I would recommend that we recommend commissioning of a study by the IOM.

MS. BERRY: I have a question for staff. Is the second bullet a second component of the same study, or is there any reason why we couldn't have -- for example, if we decided to go the IOM route, that they could not do both functions, do sort of a review of the evidence, identify gaps in the evidence, and perhaps come up with some recommendations? Do we want to have that all in the same type of report, or should it be bifurcated?

Emily?

DR. WINN-DEEN: I think we need to be clear on this, because I think you've got two different things mixed up here. One is we need to develop a guideline for establishing clinical utility. How do we know when we're there? And then it seems to me what you're talking about here is, if NHGRI or somebody is going to put out an RFA to address gaps, it's going to be for a specific disease area, a specific test where we need more evidence. So we need the generic framework. What steps should one take to collect evidence to demonstrate clinical utility? That's step one, generic guideline. And then the next one is for each new thing that comes along, how do we assure that the right evidence is gathered?

So the NCCLS document, which I think you're referring to, the guidance document that's under preparation on establishing clinical utility, belongs as part of a framework of documents that would be there as guidances to the community on how to establish clinical utility, and then the other things are specific test-by-test evidence, gathering evidence test by test. So I think it's a little bit mixed up the way it's divided here.

MS. BERRY: I think it is, too. Maybe to help staff with this, my recollection of the discussion was that there was this overarching need for a review of existing evidence, and then also to help guide future efforts by helping, whether it's manufacturers or providers, come up with or gather the correct information so that they can make their case about coverage. So this whole evidence issue is kind of an overarching theme.

The second part that staff has outlined for us I think deals more with there are going to be gaps in our current evidence base, but that doesn't mean that certain tests should not be covered or certain services should not be covered right now. What are the criteria that should be applied, whether it's a private insurer or a federal health program, in determining whether something should be covered? That's the clinical utility, clinical validity. I think a lot of that was done by SACGT. If it was done, it predated me.

DR. WINN-DEEN: SACGT went through a whole scenario of trying to figure out how to classify tests into low risk and high risk and how to make a framework that FDA might be able to use, but I'm not sure it really addressed this kind of -- it was like which things need the most regulatory oversight framework, rather than how to develop evidence of clinical utility framework.

MS. BERRY: Muin told us about some of the efforts underway at CDC. I think the point here is that for coming up with criteria, either with evidence or in the absence of sufficient evidence, we don't need to reinvent the wheel, because there are organizations out there that are looking at this, and we need to just inform ourselves. As I remember, Muin, you had the wheel and you had some pretty good information about the work that you're already doing in that.

DR. KHOURY: I think this went pretty fast this morning. What SACGT did was to develop a framework for the evaluation of genetic tests as they move from research to practice using the acronym ACE, analytic validity, clinical validity, clinical utility, and the ELSI issues. Yes, the intent was

to promote the oversight and perhaps push the FDA in the direction of incorporating this kind of acronym in the way they evaluate tests.

But the SACGT also recommended sort of a three-pronged approach. One is an FDA process, two is a CLIA process, and a third is more of a non-regulatory public/private sort of data collection process that we have been trying to work with for a long time. What we did with ACE was to flesh this out a bit more. So we took the four kind of broad evaluation of tests and developed this into a full-blown methodology. So for each acronym, there are many questions that go under that, under the analytic validity, under the clinical validity, and overall there's probably somewhere between 40 and 50 specific questions you'd like to ask of any specific test by intended use.

That work is now kind of winding down. We applied it, with the help of the Foundation for Blood Research, to five genetic tests. BRCA1 was one of them, Factor V Leiden, hemochromatosis, cystic fibrosis, and colorectal cancer testing. What we're doing right now is sort of taking stock of that experimental effort and trying to move into the next phase, working with the other agencies, AHRQ, CMS, NIH, to try to develop this next phase of a framework that uses the evidence-based methodologies that had been developed by the ACE group, but there are so many other technology assessment groups out there, including Blue Cross/Blue Shield has their own, AHRQ has a methodology, the U.K. has technology assessment, Canada has one.

We're going to be convening a group by the end of probably this calendar year hopefully to come up with a consensus way of evaluating genetic tests, again not as an exceptionalism concept, but there are many nuances there that merit maybe a special look for genetics, and then try to move with the implementation of this EGAPP proposal that I mentioned this morning.

So I think these efforts are going to be hopefully pushing us along. I think what the Secretary needs to hear from this group is sort of a need for different kinds of activities to be implemented and coordinated by the various agencies that are under his jurisdiction, because no single agency alone will be able to move this. We're not looking at the oversight regulation concept but the concept of how we can develop an evidence base, working together with academia, the professional organizations, and then put that in play in the real world so that when a new test comes along, you can evaluate it and you can hopefully guide the integration of that test into practice.

MS. BERRY: Kay?

DR. FELIX-AARON: Thank you, Muin, but I think it doesn't address sort of the gap. I mean, I recognize the gap between what Muin describes and where we are in the sense that what I heard yesterday was the need to evaluate the state of the evidence, the current state of the evidence. I think what you described is a wonderful way of evaluating new tests coming into the process. But where we are currently, I think that what we talked about yesterday was a need to develop or to produce a state of the evidence as it relates to genetic tests and services. So that's where I saw the work of the committee was yesterday.

I see up here you've described a way to develop, that what we're proposing is to develop a process for assessing what evidence base is sufficient. I see there's some interface with what Muin is saying, but there's still a gap that currently needs to be addressed so that CMS and other payers do what they need to do in terms of covering services. So that's one point.

The other point I'd like to make in terms of which organization would be best suited to do that, whether it would be an IOM study or, say, another agency like AHRQ that does this type of work. I think there are tradeoffs and there are advantages. IOM clearly has a lot of visibility, and it would definitely raise the issue. They have the credibility. But I think, though, the downside of that or the other challenges with that, with IOM, are issues around funding and who would fund that report, and the advantages for having an organization like AHRQ do this work is that it does have credibility within HHS, as well as outside the federal agencies. There's already a mechanism and co-funding for that kind

of work.

So I think we have two good options, and I think this committee would have to decide which option meets its need today.

MS. BERRY: Does anyone have a comment on that in terms of which organization? Am I correct in assuming that we're all in agreement that we need to get to the first part, which is to do an assessment of the current state of the evidence that exists today? The question is who should do that.

DR. WINN-DEEN: No, I think that's completely dependent on each and every test. For each test, you have to say what is the evidence for that test. There's no way to do an across-the-board what is the evidence for genetic testing kind of a study. I think that would be just a complete waste. What we want is how do we know when something new that comes along is ready to be integrated into medicine, and what's the continuum, what are the steps you have to take to prove that it's ready.

MS. BERRY: I heard yesterday, though, that we need both, that you can do an assessment based on the current science that's out there, what's available and what diseases exist that could benefit by these things, but then our work isn't done. You have to then do exactly what you're talking about, which is to help provide guidance for the future as new technologies come out. But I am not the scientist here, so I defer to others.

But what I heard from yesterday was that there was some need for an assessment of what exists currently, but that may not be correct.

Debra?

DR. LEONARD: Well, I'd like to just reemphasize what Emily said, which is that we're talking about two different things. One is setting up general guidelines that a committee could use to assess whether something has moved from research to clinical utility, and then beyond, once there's clinical utility, making some recommendation that there should be coverage for that service. Those are kind of general guidelines that I think are potentially being developed by NCCLS. I don't know whether other groups are developing those guidelines.

Those are generic and would not move -- that discussion would not move any specific test to clinical utility. Then those guidelines would be applied by the top bullet group that you're talking about there on a test by test, disease by disease basis, looking at the evidence that's out there, and if the evidence is there, then saying this has clinical utility and should be covered. If there isn't, then identifying the gaps that need to be filled, and then that's the third one.

So I agree with what Emily said. This is exactly what Emily said before. There's a three-step process there, and one is identifying the general guidelines that could be used for the task force, then setting up the task force that would do test by test, disease by disease, and then identify that either there is clinical utility out there in the literature or there are gaps, and then those gaps would be put out as an RFA to through research address the gaps in the knowledge so then you could move a test to clinical utility.

MS. BERRY: Are you envisioning a task force that exists in perpetuity so that they are constantly making these assessment?

DR. LEONARD: Yes.

MS. BERRY: Because that's different from --

DR. LEONARD: Or commissioning task forces or one task force that would have a different membership depending upon what test was being addressed, because it's not necessarily professional opinion that you're looking for but just those people who are knowledgeable enough to look at the evidence that's out there and say it's good enough or not. So it would be probably some constant people who can generally do scientific assessments, but then also bringing in other experts who could provide additional information that relates specifically to that test.

But it wouldn't be a task force that exists for six months and then goes away, because

this is going to be an ongoing process for every new test, service, whatever, that comes along.

MS. BERRY: Now you're talking about a federally -- I don't want to use the word chartered, but for lack of a better term, a federally mandated task force that exists? Because how in this case, going back to our genetic exceptionalism, how does this differ, or why would genetic technologies require this kink of federal structure when other services and technologies don't? I mean, each individual insurer can have its own assessment task force, and they don't answer to a federal task force.

DR. LEONARD: I don't know that it necessarily has to be federal. In fact, there's a policy option that's recommended in the document that says that CAP or other professional organizations could provide clinical utility guidelines that would basically drive coverage decisions. So it may not have to be a federal organization. If an organization, a professional organization would step forth to do this, they may need financial support or other resources or something to be able to accomplish that.

I don't mean to monopolize this discussion.

MS. BERRY: No, this is useful, because I have a completely different recollection of what I thought I was hearing yesterday from what you're just articulating. So it's important for, please, everyone to speak up, because this gets to the heart of what we're going to be recommending.

DR. WINN-DEEN: So, Cynthia, maybe the place that would be useful to commission a state of the state kind of thing is in terms of things, what are tests that are actually covered by both Medicare in a routine way, as well as if we can get information on private insurance in a routine way. What are the ones where there's inconsistencies? You know, some do, some don't. Why is there a difference? And then there will be a whole bunch that nobody covers, primarily because there's not enough evidence yet.

But looking at the ones that have established sort of unified coverage, what criteria did they meet? How did they get to that point of having unified coverage? What is the points of disagreement for the ones that have spotty coverage in terms of a gap analysis? So why do some groups cover, some groups don't? That might inform us in terms of trying to understand what the gaps are and how to create a framework that is very clear on what all the pieces of information that one needs to have in order to get a coverage decision.

Then we also would need the third part of that, which is a commitment that once a test reaches that point and you have those points of evidence, that we don't have another endless debate about it, that it's sort of accepted that that is the criteria and there's buy-in for that.

So I think that's actually quite a lengthy thing to try and undertake and get consensus on, but it would be extremely useful. The thing that's different about genetic tests is just that there are so many of them coming along. There really aren't that many new serum markers for heart attack risk or whatever. There's one every couple of years, whereas in genetics we've just got a steady stream of things coming along, not that we're exceptional because it's genetics, but just because of the sheer volume of things that are going to be coming through the pipeline, in my opinion.

DR. LEONARD: I think it has to be some body that is accepted by all coverage entities such that you don't have an iterative process that just goes on and on and on with each. So Reed was saying yesterday that USPSTF is something that insurers buy into what they say, that it does influence greatly the coverage decisions that are made by insurers. Is that correct? I mean, did I hear that yesterday?

DR. McCABE: Yes, you did.

DR. LEONARD: But then I also heard that that process is also very slow and takes forever. So can we get an equivalent body that moves more quickly, or is the process just that slow?

DR. McCABE: I think that body, it's not only slow, it's also incredibly rigorous. If we held medical practice to those standards, we would find that we were doing very little. So I think part of the discussion yesterday was that we need to develop some process, but perhaps we have to have a

process that's really workable and will bring more of these tests and services into practice.

DR. LEONARD: But without that end buy-in, and I don't know how you get that up front, but without that final buy-in, the whole process may not be that useful to invest in if it's not really going to influence many, not necessarily all but many of the different insurers that are out there.

MS. BERRY: Kay, and then Muin.

DR. FELIX-AARON: The U.S. Preventive Services Task Force approach would be a test by test, a service by service strategy, and not addressing what Emily said earlier about what is the state, because I think we came to this question as what was the problem. The problem was that those types of services aren't being covered, and that's how we defined the problem. So we said they're not being covered because there's no evidence.

So the question is how would this group want to proceed? Is this group going to proceed by looking at every service that is available now and looking at what the evidence is, or pretty much trying to address what Emily says? There are lots of services coming on board at this point in time, sort of circumscribing the most important services or the most promising tests and seeing what is the evidence for the benefit of those tests for the public, because I think it would be a higher level analysis than a test by test analysis, where you look at a specific test, but looking at the body of this information.

I mean, does it warrant departmental action at this point? I mean, is the body of evidence enough to say that there needs to be some statement about what purchasers should be covering? Because this is a new area, it's a reality, and they should be focusing on genetic testing and services.

MS. BERRY: But can you make an assessment like that of a potential technology that's coming to the fore but hasn't come yet? There's no evidence. You don't really have anything to assess at that point if it hasn't really come out yet. So we're sort of in this limbo land. There are evidentiary gaps, I think, with regard to existing technologies, but then we recognize that as new technologies come out and are developed, we need to provide some guidance. I think this is what I heard others say, we need to provide some useful guidance so that we don't have to go through this over and over again, going back into the literature. Instead, there will be guidance on the front end so that those who provides these services or tests or technologies will have that information, and that will inform coverage decisions more instantly than currently happens.

DR. FELIX-AARON: I hear what you're saying, and I think the group will have to make that decision. What I've heard is that there's a compelling argument to guide the process going forward, but I think that doesn't take away the responsibility for purchasers and other groups to assess the evidence, because clinical trials -- people will have specific questions that they're asking for a particular study, and there will still need to be somebody looking at issues of benefit, looking at effectiveness, and making those types of comparisons.

So I think the guidance is clearly important as we move forward, but I don't think it will remove the need for the work on the back end saying should we cover this or not, the decision-making processes that purchasers have to make.

MS. BERRY: Muin, did you have comments on this?

DR. KHOURY: There are lots of issues that are being discussed, and I think sometimes we mix apples and oranges and pears. I think while there may be a need for a general assessment of the status of the state, where we are with genetic testing, I think we've heard enough over the last few years that we need a rigorous methodology to begin to look at the validity and utility of genetic tests by intended use, test by test. So not to negate what you just said, Kaytura, but to sort of move it along the test by test methodology.

The process which I described this morning, which I probably did not describe in any reasonable way with that fancy diagram in there, will take us a long way to try to begin to bridge that gap between where we are today and where we need to be in the future. The experiment we did with the ACE

project, especially interacting with the U.S. Preventive Services Task Force, I'll take you through that for a minute.

We've established the ACE framework with very detailed questions, and then we funded one of the AHRQ evidence-based centers, the Oregon one, to look at the BRCA1 testing. They are working through the methodology. The U.S. Preventive Services Task Force uses a very rigorous methodology that primarily focuses on clinical utility. It doesn't deal a lot with the ethical issues, or even the analytic validity of the test. What we were told, and we have an ongoing discussion right now with the evidence-based centers and with AHRQ, is that for most of these new genetic tests, the return will be insufficient evidence from that AHRQ, very detailed, rigorous look.

So after this initial phase of trying to put all the technology assessment pieces together, because different organizations have different methods of evaluating tests by intended use, the plan is to put together a working group. We didn't want to call it a task force because we did not want to create another U.S. Preventive Services Task Force but we want to work with the existing one, to create an independent working group that's really not CDC owned or NIH owned or AHRQ owned.

But basically, they will begin to look, test by test, they will decide for themselves, guided by the horizon scan and the stakeholders, first arriving at a consensus for test methodology review, and then review test by test the whole spectrum, from analytic validity to the ELSI, make some pronouncement of what we know and what we don't know through using evidence-based centers, probably using the AHRQ evidence-based center reviews, putting those on websites to try to influence interim policy, because many of them would return insufficient knowledge, lots of gaps, and then working with NIH and others to fund the various research that needs to fill that gap.

I'm trying to follow Debra's comments. All you said here is sort of what this process will move forward to. Again, not one institution, one organization will be able to do this alone. It has to be sort of a joint public/private partnership. Forgive me for keep singing that same tune which, Ed, you probably are tired of me over the last 5 to 10 years, but I view this as an essential way of moving forward, supplementing all these various processes that already exist within HHS and the FDA and CLIA, et cetera.

So I think what you have begun to articulate there is essentially that vision that the diagram I presented this morning tries to move us in that direction, and maybe what we need to do is spend some more concentrated time, maybe the next time or the time after, to flesh this out in a way that engages all the stakeholders, because this is where the rubber meets the road. This is probably the most important thing that will drive the true translation of genetic technologies into practice. I mean, education is important, but without the evidence, there is nothing to integrate. So I do feel passionately and strongly about that, and we will continue to work with our sister agencies on this.

MS. BERRY: Muin, do you see any value in having IOM or somebody else do this first component, or is that not necessary given how far along your task force is moving?

DR. KHOURY: I think the IOM has a wonderful utility. As a matter of fact, after this meeting today, I'm going to the IOM tomorrow. The Disease Prevention and Health Promotion Board is having a meeting to talk about genetics and public health, a special sub-group to evaluate where we are in that process, and they will have another meeting in September. I know NIH at one point talked to them about a review of the cohort studies and the concept of a cohort study. I don't think that dialogue has yielded some result.

I think if we want to approach the IOM to develop an IOM report, which is a full-blown picture, it has to be well thought out, and it has to be kind of a broad mandate, because the IOM pronouncements take time, they are not cheap to implement, but they have a lot of weight. So if you want to go to the IOM, I would encourage you to think about it, and maybe the feds can sort of talk among themselves in terms of if there is a unifying agenda along that translation pathway that would necessitate

a full IOM review. I think that would be a great thing, but it will take time and very deliberate discussion before we go to the IOM.

MS. BERRY: Debra, and then Emily.

DR. LEONARD: So it sounds like you are already fairly far down the road in the planning part of this work group that would do the test by test evaluation using the ACE process, is my understanding.

DR. KHOURY: ACE-plus, which means merging the best tools of the trade, which would be ACE plus the other technologies that are used by the U.S. Preventive Services Task Force, the Cochrane Collaboration, the Canada Technology Assessment. There are lots of groups out there that do this stuff.

DR. LEONARD: So that first part, when we had turned that two-step process into a three-step process, that first step is basically done, or at least in a workable enough form that you could start doing test by test evaluations once you can reach a consensus among all the participants as to what the categories of evaluations are going to be.

So how can we assist this work group in happening and moving forward? Do you need funding? Do you need a commission? How could we move that process along or make a recommendation to the Secretary that would assist in facilitating this process?

DR. KHOURY: I think SACGHS could be a wonderful voice with the Secretary to kind of stress the fact that this process needs to happen, and it has to be a collaborative process across all the agencies that are under the HHS Secretary, and that involves the private sector as well. So promoting the concept of a public/private partnership.

At this point, you can make that assessment and then follow and be engaged in the review of how far along will this process really go forward to fill the gaps that need to be filled. So I think by being engaged, by communicating with the Secretary about the importance of this process, about the collaborative nature of it, I think that would be sufficient at this point.

MS. BERRY: Emily?

DR. WINN-DEEN: So I was going to ask if you'd be willing to give us either a written briefing by forwarding on the materials that you already have between now and our next meeting, or if you don't feel they're quite ready for that, to give us a briefing at the next meeting that's really much more in-depth, walk us through the questionnaire, what are all the lines of evidence, what are the questions, what are the things, and sort of where are you, who are the stakeholders that you're working with, what are their concerns. Maybe we should hear from them independently.

I really think we've identified that this is a critical activity that needs to happen, but we don't need to reinvent it if it's already happening. Well, we either need to bless what you're doing and throw our support behind it or make whatever suggestions that this team might have for how it might be adapted or improved, rather than trying to go through creating a whole new mechanism. I'm not in charge of the agenda for the next meeting, but if we're going to have coverage and reimbursement, I would like to put that as a potential agenda item.

DR. LEONARD: Also to explore whether or not, after you go through this process for each test, would the insurers then buy into that as evidence to use in their coverage decisions.

DR. KHOURY: That we don't know. It's part of the experiment in the next two to three years. So I think depending on when the next meeting is -- the next meeting is in October, we'll be ready to give you something in writing and maybe have more of a discussion about that.

Now remember, this process is not necessarily to drive coverage but to summarize what is known and what is not known, and then by having the right people at the table, then further discussion could lead to this leveraging of the coverage issues. I mean, the way we started this -- I'm glad you kind of separated 1 and 2 here -- is developing the evidence base. This is sort of what that process is

geared to, sort of summarize succinctly what we know and what we don't know, and where the gaps are, so that further research can be done to fill those gaps. Then in the interim, the information that is available can be used for some interim policy or guideline development. If not, then we go back to the drawing board and wait for the research to fill the gaps, and then go at it in another cycle.

So in the meantime, the problem is that people are asking for coverage in the absence of sufficient evidence, and that's something we're going to increasingly face in the world of genetics and genomics. Silence is not really an option. By being completely silent, it's basically not stepping up to the plate and saying is this a good thing or not a good thing. I mean, somebody has to step up to the plate and summarize the status of information so that both consumers and health care providers are armed with the right evidence at any given point in time.

MS. BERRY: A question for the group. We could take several different approaches. One approach would be to, in our report, in the recommendations section, we would state, of course, earlier in the body of the report the nature of the problem, and then in the recommendations section note this working group effort that's going on and talk about the need for supporting that effort and referring to it in some way. Or do we hold our report until that effort is further along, and then the report would simply endorse whatever the approach is that's taken there? I don't know if you have an opinion one way or the other on that.

Ed?

DR. McCABE: I would suggest that we not hold our report, but that we move forward, that we document whatever the state of the art is at the time that we finalize this, and I would hope we could try and finalize it at the next meeting, but we simply document where we are at that point in time.

MS. BERRY: So the recommendation could be something fairly general about the need to develop a well thought out methodology or process for evaluating the evidence and looking at all the factors that insurers or federal health programs need to look at in order to assess whether a particular technology or service is covered, and then refer to this effort as a potential model that we'll be monitoring, without coming to a firm conclusion as to what those precise criteria should be, because it sounds like that effort is under way.

Sherrie?

DR. HANS: One of the concerns that I had in the discussion yesterday and continues in the discussion today is -- and I don't know if this is the intent of the committee or not -- that you're setting up a higher standard for coverage and reimbursement for genetic tests and services than for other medical interventions and treatments. I'm not sure that that's what you want to do.

The other concern that I have is that you've taken a lot of public testimony on this already, but has the question been asked of payers what would be the most useful for them as they go about making their decisions, what is the information that they're looking for? Certainly from the VA's perspective, which is admittedly in an odd category because we're payers, providers, and a public health agency all in one, there are sort of three levels.

One is what are the things that we should tell our docs they cannot order, we will not pay for, we're not going to have folks using those technologies, and that's sort of the basic minimum of do we think this has any validity and does it have any minimal utility at all. Then there's a whole range in there where it's up to the clinical decision-making and perspective of the providers about what is appropriate for the patient that's sitting in front of them, and that's medical decision-making, medical practice that we sort of leave to our physicians.

Then there's a very high level when we use USPSTF or we say is there compelling, overwhelming evidence to say that this is something that, sort of as a public health agency with our population, that we want to ensure that we're pushing forward, that we're making sure that everybody gets

this test, that all of our docs are doing this, and that's the next level. So there's the no, there's the medical decision-making, and then there's where do we really want to put our emphasis and where is the real high level of evidence that we're really going to push around.

From just the VA's perspective, helping us understand where to draw those lines through the kind of guidelines and guidance that you've been talking about would be helpful, but I say that in the context that I would hate to see this committee set up a higher standard for this technology for coverage and reimbursement than for other medical interventions.

MS. BERRY: Emily?

DR. WINN-DEEN: I just want to quote my friend, Sam Broder: "Don't let the perfect be the enemy of the good." We need to understand at what point we have enough evidence to put it into practice, to reimburse for a new test, and that's I think what Sherrie was talking about, from no to at the physician's discretion. If the physician feels it's medically necessary, they order it, and it should be reimbursed. So we need that -- for the coverage and reimbursement purpose, that's sort of the threshold that we need to define.

Now, the next level up in genetics I would say is population screening. At what point does everybody need to have this test because it's so important? For that I completely agree, there's another level of evidence that's required to get to that point, and that's maybe to some extent the kinds of tests that CDC has been focused on, ones that are at least candidates for a population screening approach.

So I think that part of what developing the evidence base is is we have to have very clear cutoffs on what pieces of evidence have to be there in the general consensus of insurance providers for a test to be covered, and that gives everybody sort of the same bar to aim for, no matter whether it's test A, B, C or D. You know what you have to develop, you know what to expect. If people want to order it before you've reached that threshold, they know that they're not likely to get coverage.

But on the other hand, once you get to that threshold, then it shouldn't be uneven. It shouldn't matter if you're employed by the government and covered by government insurance or employed in the private sector and covered by private insurance. You should have that covered. I think those are the kind of inequities that we're trying to get past and trying to identify how to deal with that. Then the part we haven't gotten to yet is how do we get the right level of reimbursement associated with that. But if you don't say a test is worth covering, it doesn't matter.

MS. BERRY: Kay, and then if someone can volunteer to wrap up the two, because I do think there are two parts here, and we need to really home in on what will be our two recommendations under this coverage section. I think we're getting there, but I'm not positive yet.

Kay, you had a comment?

DR. FELIX-AARON: I hear what Sherrie says in terms of the description, the different bars, and I agree in terms of making those types of distinctions. But what we also hear is purchasers and people who are making decisions, they're making payment decisions or making clinical decisions, asking for guidance as to what should be covered or what services should be offered. So I think that the bar is high, but I think that it's not only for genetic services. It's for newer services as medical practice has moved more and more to a recognition, that anecdotal experience, that sort of clinical practice needs to be supplemented by rigorous evaluations of what is the best course of action, whether it be in the area of payment or whether it be in the area of what types of services should be provided to patients. We also get that response, the need for more guidance.

MS. BERRY: Anyone volunteer to summarize, then? Come on, we can do it. Just two recommendations.

All right, evidence base. Do we need a study? Do we need an assessment? The state of the state.

Debra?

DR. LEONARD: I don't think so.

MS. BERRY: No.

DR. LEONARD: I think that the first bullet should be -- I don't know whether you want to say specifically the CDC's work group that they're organizing, but develop a mechanism for assessing when the evidence base is sufficient for coverage or for establishing clinical utility or for moving it up from don't do this test to medical decision-making use of the test, and this would be done test by test.

The outcome of that, which are the sub-bullets -- you have also identified gaps, but the outcomes of that would be either that process establishes the clinical utility or it doesn't establish the clinical utility, evidence is there, and we'll identify the gaps in the evidence base and various applications, which will identify areas for research that need to be done. Then the second bullet would be that if that second step happens, that there's a mechanism by which to get an RFA out there to have that type of research done that will then take it back up through this iterative process and hopefully end up with that the clinical utility is established by the process.

Does that make sense to you guys who are writing this down?

MS. BERRY: Hunt?

DR. WILLARD: I'm confused on the potential for an RFA here. So for a particular genetic test -- I mean, this is an ongoing process that changes between Monday and Tuesday as the potential for a test is developed and different cohorts are evaluated with different odds ratios, or whatever. So I can't imagine a situation in which one would have gone through a sufficient cycle of responding to an RFA and going out there to find out that, oh, while we were waiting to do that, we got the answer a year and a half ago.

I realize there are some questions that are more complex than I just spelled out, and the utility for population screening for CF alleles is a prime example, or hemochromatosis or what have you, but to me it wouldn't be a general RFA. It would almost have to wait for a failed process where there isn't sufficient knowledge coming from the regular pipeline that we're all engaged in as new tests come out of our institutions or other institutions. It's only when there fails to be a consensus reached, perhaps because there's different populations, perhaps because there's different technologies, whatever, that one would finally get to the point of saying, gee, we need a much more concerted effort to try to see whether the answer is thumbs up or thumbs down.

MS. BERRY: Ed?

DR. McCABE: That's where I would see it. I wouldn't see it as a blanket RFA for all genetic tests. I would see it as a targeted RFA when tests with potentially high value, gaps were identified, it's not clear that they're proceeding down the road toward implementation, and hemochromatosis is an obvious example, where you need large population studies to carry that out and determine the penetrance of the various alleles and that sort of thing.

So I see this as not a blanket RFA for all genetic tests but certainly a prioritization, identification of those that could impact heavily on the public's health.

MS. BERRY: Muin?

DR. KHOURY: Actually, I was going to use the example of hemochromatosis as the poster child of this process. Hemochromatosis happened at the time -- I mean, the gene was discovered in 1996. There was a rush to pronouncement that we need population screening. We and NHGRI put together sort of a working group, an expert panel that essentially looked at the evidence. I mean, we didn't do it in this AHRQ type method but just convened the expert panel and looked at things, and then decided that there was not enough evidence, and the research gap was what is the penetrance of the hemochromatosis with respect to various health outcomes.

A feedback group went back to NHGRI and I think NHLBI on this, and they funded

this gigantic cohort of 100,000 people to begin to look at the natural history of the hemochromatosis gene. Now, what we need to do here -- I mean, the reason why this is important is because we will have many such applications. As time moves forward, we may be hit with two or three similar claims every week. It hasn't happened, so this is a good time for us to plan for it, and I guess always the value added of any process would be why not leave it to the existing mechanisms.

I think this group and other groups have decided that genetics may put a pressure on the system because just the magnitude of the quantity of genetic tests that may be hitting us in the next decade or two may overwhelm the system and the ability of evidence-based groups like AHRQ, U.S. Preventive Services Task Force to cope with it.

So I think the hemochromatosis model is a good example, and you multiply that two or three or four or ten times and you begin to develop that kind of genetic process that examines the evidence using specific quantifiable methodologies, identifies the gaps for that particular test, goes back to the research and the feedback loop while more knowledge is accumulated, while at the same time communicating in a transparent way what we know and what we don't know so that the right coverage decisions are made, the public is more informed, and the health care providers are more informed so there is a feedback loop to everyone.

DR. McCABE: I just wanted to let everyone know what I asked Cindy before, and that is that we try and wrap this up by quarter of the hour so that we have an hour left to deal with what we had left about two and a half hours for in the schedule. I know that will be pushing it, but if we can try and give recommendations to staff to help with the redrafting of this.

MS. BERRY: So we're abandoning the notion of a state of the state study. Do we have consensus there? We're not doing that.

Hunt?

DR. WILLARD: I would agree with that point, but following on Muin, two comments for staff. One is, when in doubt, keep writing, in the case of a particular test or in the case of a particular association between an allele and a clinical outcome to be sure we're not talking about general, one-size-fits-all for all genetic tests but that it relates to a specific one.

Then in the report, I think the case story of hemochromatosis is a wonderful one to put actually into the report itself, because it does demonstrate if there was anything that looked like it should have been a slam dunk, that was probably it. Of course, it turns out to be very, very different from that. So it does illustrate exactly how this all may play out for 100 other tests.

DR. McCABE: It turned out to be a dunk rather than a slam.

DR. GUTTMACHER: Can I just emphasize what Hunt said at the end of that, and that is what it means for 100 other tests, because I think as we move forward we can't believe there's going to be any kind of a mechanism that can actually vet every single genetic test that comes down the line. So part of the interest in hemochromatosis I think was using that as a paradigm for a certain kind of genetic testing. I think we need to look at those particularly. We need to think of ways of developing paradigms that we can then use, because as Hunt also brought up, any genetic test, the use of it will change over the course of a couple of months in different populations, all kinds of issues.

So there's no body that one could have, particularly in the current American medical system, that would be able to sort of vet each test, and we shouldn't ask for that. Instead, we should look at ways that we vet processes and ways of thinking about things so that they be used wisely.

MS. BERRY: Suzanne, Amanda, do you all have enough information based on the discussion about the working group and their efforts, and maybe sort of the nuggets of a possible recommendation for inclusion in our report regarding the criteria?

MS. SARATA: Just one quick question for Debra. Could you clarify, when you say develop a mechanism for assessing when the evidence base is sufficient, did we decide who was going to

be responsible for doing that? Was it a federal agency, interagency --

DR. LEONARD: (Inaudible.)

MS. SARATA: But you said not to refer directly to the --

DR. LEONARD: Well, that's up to the group whether you want to do that. But we haven't heard about that in detail. We heard about it at this meeting, so it's a matter of -- I would refer to that process as something that the Secretary could enhance, facilitate, support, once we've heard about it in more detail and know that that would be a mechanism. But it sounds, at least from what we've heard, that it would be a mechanism to do what we're asking to be done. So I don't know what everybody else thinks about mentioning the CDC. If they present at the next meeting, and this report is going to be finalized at the next meeting, then we could put it in temporarily, and if we disagree with that once we've heard the CDC's presentation on the work group, take it out or change it.

MS. BERRY: Muin, do you envision the model or the methodology that you all are going through in the working group as something that once it's finalized, however long that takes, it could serve as a model for private insurers, or is this something that only some sort of federal task force, group, entity could undertake? Is it translatable into the private sector?

DR. KHOURY: Yes, potentially. I think the best way to characterize this process, as my friend Elliott Hillback from Genzyme always said, it is an iterative process. We've iterated for the last few years. We've reached a point that we are closing in on the methodologies for the review. I mean, that's step one, because whatever group you basically form has to be armed with a set of methodologies so that if you form another group, they can come up with the same conclusions because of the idiosyncracies of the system.

The second is now the experiment over the next two to three years is to test the feasibility of this approach. Alan mentioned, and other groups, that there are existing other processes, and what we need to do is test whether a process like this might work in the current set-up of our health care delivery system, given that there is Medicare, Medicaid, private sector, et cetera. So by constructing very carefully a process that brings all the partners to the table, and evaluating it, because part of the experiment is an evaluation component, within three years we'll know whether this is a model to implement and sustain, or not to implement or sustain.

We're in the beginning process of Phase II. Phase I was the development of the methodologies, and we're finishing with that. Phase II is the development of a model process to see whether it will work, and then package it in a way that fits with the existing processes that we have under the medical system right now. So within three years we'll have an answer, but within a year we'll know whether at least -- I mean, you'll be hearing more of the attributes of how that works, and this group can really weigh in in a big way to steer it one way or another as the experiment unfolds, I think.

MS. BERRY: So Amanda, what I'm hearing, then, is that I don't think we necessarily want at this point to recommend that there be some sort of federal entity or structure for evaluating all genetic tests as they come along, but rather that we are taking a good, hard look at this approach that's underway and we'll be evaluating it as it progresses, with a view towards determining if it's a model that can be used across all federal health programs and in the private sector and elsewhere.

Debra?

DR. LEONARD: So moving on to number 2 at the bottom, I don't think it's so much develop criteria for coverage, because that's kind of what you're doing in 1.

MS. BERRY: Right.

DR. LEONARD: So really what we want to do in 2 is facilitate the use of the evidence base as criteria for coverage. So it's basically using what's in 1 or facilitating the use of that by CMS, which we do. The Secretary of Health and Human Services does have influence over CMS. But then also to explore whether other insurers would use this evidence base in their coverage decisions, and I

don't know a mechanism for doing that. It can directly be done with CMS by some mechanism that could be developed, but what you do for other insurers -- but we've also heard that what CMS does influences what other insurers do. So that may be a way to have an influence in and of itself.

MS. BERRY: Also in that number 2, I'd actually move the last bullet that's on the last page of the outline that you have, where it's overarching barriers, the preventive nature of the genetic tests or technology. That actually could be moved up into the coverage section because we heard a lot of discussion yesterday about one of the barriers to coverage in Medicare is the fact that there is a statutory exclusion with regard to screening tests and services. So there was discussion about legislation that's been introduced or is about to be introduced in Congress that would allow Medicare to cover certain preventive technologies.

So I think maybe, unless folks disagree, that could be a part of our coverage recommendations. We could talk about the screening exclusion, that perhaps that should be changed. That would require, of course, a legislative change. It's not something the Secretary could do unilaterally, and we of course can't lobby Congress to do it, but it could be something that we reference in the report that the Secretary could focus on and, as mentioned here, make reference in the administration's submission to Congress, budgetary submission.

Ed?

DR. McCABE: I would suggest that perhaps we could deal with that last page by that recommendation, and then we had already, I thought, whether now 2 and 3, provider education and training and health disparities, that we intended merely to make those as paragraphs to elucidate the problem in the body of it. I thought that was where the discussion was yesterday. So we had removed them from recommendations per se and made them just something we would reference as they related to coverage and reimbursement and not in the grand scheme of the education and training or health disparities.

MS. BERRY: That's my understanding.

DR. McCABE: So that leaves us with reimbursement to cover in the next three minutes.

MS. BERRY: Debra?

DR. LEONARD: Can I take one of those minutes? What is not on here is the CPT modifier system will reduce denials but there's still the issue that's not listed anywhere on here, which is the inadequacy of the level of reimbursement for the cost of doing these technologies. So there are royalty fees, but just the reimbursement level for the CPT codes that do exist is not adequate for the cost of doing the tests, in general.

MS. GOODWIN: Can I prompt you a little further and ask you what do you recommend as a mechanism for making changes?

DR. LEONARD: Like I said yesterday, the whole reimbursement for CPT codes issue is very complex, and when you raise the reimbursement for one CPT code, they kind of want to reduce the amount paid for other CPT codes. I don't know how you hit a balance, but if we are going to move toward genomic medicine, where this genetic technology-based testing is going to be used more and more and more, it's not a viable system as it currently exists because the payment is inadequate to cover the cost. That's an issue that has to be addressed if we're going to move forward.

I don't know within the current reimbursement for CPT code, establishing that reimbursement level, how you do that, because I've heard that it may end up opening up the entire laboratory fee schedule for review, and I'm not sure that pathologists -- I mean, I may be dead as soon as I walk out of this room if that is the conclusion of this discussion. So it's a very complex issue, and I don't know how you change that.

So one way, maybe when we're doing the discussion next time, is to have someone

come and inform us of how you do changes in reimbursement.

DR. McCABE: That would be someone from the AMA and the group that's involved in that process. We could identify the appropriate individuals.

MS. GOODWIN: You're talking, though, about changes to the actual coding development process?

DR. LEONARD: No, the codes exist, although there's discussion of bringing online new codes. But that process happens. It's the codes, the 14 codes that we currently have where, when you put those together in combinations, the amount paid by Medicare for those services does not cover the cost of doing that test.

MS. GOODWIN: So perhaps AMA is not the right group, then, to bring here?

DR. LEONARD: Well, it's AMA that establishes the reimbursement level, but those reimbursement levels were established back in 1980-something when these codes were developed and there wasn't a good idea of how much it costs to do this testing. So that's the reimbursement level that still exists for those codes.

MS. GOODWIN: My understanding is, though, that AMA doesn't set the reimbursement level. They simply set up the framework for the codes, but the AMA is not involved in actually determining the payment rate associated with each CPT code.

DR. LEONARD: Well, Mark Synovec from CAP deals with this all the time, and I know he understands the process, but I have yet to absorb that into my brain. You're right, the AMA is the one that establishes the new codes, but then there's a complex process of collecting information about the time and the cost and the professional components and technical components of everything to establish reimbursement.

We've gone over our three minutes.

MS. BERRY: Hunt, did you have something?

DR. WILLARD: I was only going to ask the question, and I feel your pain, Debra, but to what extent is any of this specific to a genetic test as opposed to the introduction of all kinds of new tests, including putting in a variety of medical devices? I mean, there must be all kinds of groups that say we're not being reimbursed adequately for what we've put into this. So I would caution against -- unless we can identify that there are specific issues related to genetic testing, I would caution against sort of railing against the CPT reimbursement system because I don't think that's going to get us very far and we'll undercut some of our credibility, unless we can specifically target it where there are inadequacies to how they're dealing with genetic tests as opposed to four other classes of tests.

DR. LEONARD: I just know that I do genetic testing and all kinds of molecular testing in my laboratory, and I'm very often faced with the issue that we want to bill another institution, because if we bill insurers, we don't get paid, and yet the other institution says they won't send us because they're going to eat the cost. But they're perfectly happy to send it to my lab and have my institution eat the cost of doing this testing. So there is something inadequate in the system for molecular tests because they are not broadly available everywhere, like CBCs and other laboratory tests. If there's inadequate reimbursement, there is inadequate reimbursement, it's not evenly distributed across the health care system.

So if we're going to be moving more and more in that direction of doing more and more genetic tests, the system has to be fixed. It may be that there are problems in radiology and there are problems in other specialty areas. I don't know those, but I am acutely aware in my laboratory that there are discussions that go on about people not wanting to be billed for the test because they know that they aren't going to get paid, and yet they need the test because it's standard of care.

MS. BERRY: Ed?

DR. McCABE: Can I ask for some other big questions? And then we'll have to give

it back to staff and the committee to work on in the interval between now and the next meeting.

MS. BERRY: Emily?

DR. WINN-DEEN: I just wanted to see if we could get a couple of words into the reimbursement system relating to health economic value of tests rather than cost reimbursement or under-cost reimbursement. I think if we're trying to frame what are the key issues, I think those are the key issues. Not only are we not recognizing the value that these tests have to the overall management of the patient, that is where in some cases they save a whole lot of money in hospital stays or other places, but we also need to recognize that many of them are under-reimbursed, and we should just frame those as issues.

MS. BERRY: Judy, I don't know if you can help us with this, or someone else who might know. If somebody wanted to influence the level of reimbursement for a particular CPT code, how do you do that? I mean, is there a recommendation that we can come up with, is there something that's missing that currently does not exist that results in this inadequate level of reimbursement and that we could recommend something to the Secretary to provide that missing piece that would help increase rates? What is the problem there? I'm not familiar enough with how the reimbursement is actually set for individual codes.

Ed?

DR. McCABE: As I've been taking notes about possibilities for the next meeting, I have down that we should educate ourselves about the process of establishing CPT code reimbursement.

DR. WILLARD: Didn't we do that?

DR. McCABE: No.

DR. WILLARD: We had somebody come talk to us about that. He was adding up the cost of a Southern blot and a PCR and everything.

DR. McCABE: Yes, but this has to do with how you influence the system, because that's the way people do it now in order to get increased reimbursement, but it's not how you influence the system to change the system.

Can I ask that if there aren't any other big issues, I want to be sure that we have time to deal with the education and training. I think this is a huge undertaking that we've begun here. I appreciate very much Cindy and her group's efforts on this behalf. But I think we're close to completing the resolution on genetic education and training of health professionals. I want to be sure we have time to look that over one more time before we go on to planning the next meeting and other issues.

Joan?

DR. REEDE: I think everyone has a copy, a hard copy of what was previously read to you. I don't want to go back through reading it again.

Have you had an opportunity to look at it?

DR. McCABE: Why don't we give everybody three minutes to read through it. It's not a terribly long document, so you can read it through probably faster than if we read it aloud for you.

Is there anyone who doesn't have this hard copy before you?

(No response.)

DR. REEDE: I'm assuming we've had our three minutes time to review. If we could take them a page at a time, are there any comments or thoughts in terms of the first page of whereases?

DR. TURNER: Number 6.

DR. McCABE: I'm sorry, we lost your mike. If you could put it on again.

DR. TURNER: I'm sorry. Number 6, to assure equitable access, I'm not sure that we're not overstating it to say that education of health care and public health care professionals is necessary to assure equitable access. It's a piece of it.

DR. REEDE: How would you like to change that? "Is a necessary component to

assure"?

DR. TURNER: Yes. It's a piece of it.

DR. REEDE: Add the word "component."

DR. TURNER: Yes, I think that would work fine.

DR. REEDE: Other comments?

(No response.)

DR. REEDE: If we could move to page 2, comments or thoughts?

DR. WILLARD: Joan?

DR. REEDE: Yes?

DR. WILLARD: In the last of the bullets, oddly enough, the word "genomics" is singular and not plural. So it should be "genomics, its ethical, legal and social implications."

DR. REEDE: Thank you.

Others? Debra?

DR. LEONARD: In reading through the bullets, I think on the second page, not the bullets at the top but the little paragraphs at the bottom, "Promote and support initiatives that address the integration of genomics into education and training of all health professionals," that's really the same or a component of the next bullet. So you could say, "In order to promote, support, and facilitate the integration of genomics into health care and public health now, direct HHS." So those two things I think could be combined into one.

DR. REEDE: So what I'm hearing is that removing the third recommendation and incorporating it into number 4. "In order to promote, support, and facilitate the integration of genomics," et cetera.

DR. KHOURY: Since we are one Department, I guess we don't need HHS to direct us. What I would recommend is that you would say that for HHS to develop a plan to support the development cataloguing dissemination of blah blah blah case studies. So basically we leave it up to our Secretary to decide how he's going to work with the agencies, rather than just direct us to work in a fragmented way but develop a plan for how the Department will do this as one entity.

DR. REEDE: Fine.

DR. HANS: I'm not sure, Debra, that those two actually are the same. I mean, one is the --

DR. LEONARD: Right, because I realized that the last one goes on to talk about developing case studies.

DR. HANS: Right, cataloguing and disseminating case studies. The other is a more broad, general, integrating into.

DR. LEONARD: Right. I stand corrected.

DR. REEDE: So given that we said two different things, are we keeping them separate? Is it the committee's wish to keep them separate?

DR. LEONARD: Yes, keep them separate.

DR. REEDE: Fine.

MS. CARR: I'm sorry to go back to the bullets, but in the first one and the last one, we're only using genomics, and I'm wondering if we need to add genetics and genomics in both those places, and then it becomes "their," not "its," I guess.

DR. REEDE: I think in most of the document we've used genetics and genomics, and I think to be consistent we could use the same language here.

DR. FEETHAM: That's also true of being consistent with using health care and public health professionals wherever that belongs.

On the second page in the larger paragraph or divider, using the term

"genetics/genomics revolution," I think we're beyond that language, something I've learned from Alan and others quite a while ago, that maybe it's the benefits of genetics/genomics knowledge, not having to say revolution.

DR. REEDE: Fine.

DR. GUTTMACHER: I'd like to volunteer the most picayune comment, and that is down at the bottom, the listing of organizations, it's actually the Coalition for Health Professionals rather than of Health Professional Education in Genetics. Just to show you I was paying attention.

DR. REEDE: I think the other part is there are some others that responded either in oral or written testimony that are not here, so we'll be correcting that and listing them alphabetically and all of those types of things.

If there are no other comments on page 2, could we turn to page 3?

DR. WILLARD: The two that begin at the top, provide and promote, those two seem very, very similar to me, and there must be a way to combine those and save 45 percent of the words, because they seem very repetitive to me. One is training for the implementation of models, and then the next one is promote communication to enhance dissemination of those models. There's not a lot of difference there to me.

DR. REEDE: Do you want to suggest language?

DR. WILLARD: I have infinite respect for the staff to be able to merge those two somehow.

DR. LEONARD: Or you could simply say "for faculty training and the implementation and dissemination of clinical application-based genomic," because basically you just want to get them out there, which is the point of the second one.

DR. REEDE: And I think also the first one, a major part of that was the faculty training, because that was a recurrent theme. So we'll leave it to staff to wordsmith that.

DR. WILLARD: Also, the one that refers to pipeline programs, first of all, it isn't clear to me at least. Maybe the Secretary knows what that is. But second of all, that didn't seem too different than the third paragraph on the previous page, "promote and support initiatives that address the integration of genomics into the education and training of all health professionals." Isn't that the sense of this one, to support training programs?

DR. REEDE: I think pipeline programs does not refer necessarily to health professional training but actually to the programs that may be K-12 or college or other programs that would bring people into the health professions and provide this background. So the pipeline programs refers to a different level in the academic pipeline.

DR. WILLARD: That's very helpful, but then I would suggest we spell it out and not call them pipeline programs, or put parentheses, K-12 programs or something like that.

DR. McCABE: Why don't we call it inter-educational K through 12 and undergraduate pipeline programs?

DR. REEDE: Fine.

Debra?

DR. LEONARD: So I hate to add something, but it was brought to our attention several times that many of the competency recommendations are that generalists be able to identify when they have a genetics issue or one that would need specific genetic counseling, and that the genetic counselors are gearing up to deal with more complex disease traits, et cetera, and that there are a paucity of genetic counselors that will likely be insufficient for the growing need. Do we want to make a recommendation, as we've been requested to, about specific genetics training programs in addition to the general programs, which is what all these different recommendations really deal with?

I know Muin has some concern as to whether or not we will need genetic counselors,

but I cannot believe that interpretation of the genetics or genomics of complex disease traits is going to be any simpler, really, than the genetics of single-gene diseases, and I think we still will need specialists, and we may need expanded specialists who can deal with these, especially the whole time aspect of doing genetic counseling with family members that most general practitioners don't have the time to do or the expertise.

So that's something that has been brought to this committee by different groups, and I don't think we've addressed it in this resolution.

DR. REEDE: I think part of our deliberations, this didn't come up as something to include yesterday, but we also determined not to include anything for specific disciplines. So one of the things that came back was that we need more nurses in order to be able to deal with these issues, and we thought that if we started to deal with specific disciplines as in nursing, as in genetic counselors and others, that they may take the form of another kind of study or recommendation, as opposed to this, which is really looking across the full spectrum of the health professionals. So most of this and most of the comments that we got back really pointed to commonalities across the full spectrum of health professionals that needed to be addressed, and so that's what we tried to incorporate here, with an understanding that we may need to come back and look at specific disciplines in terms of issues.

MS. ZELLMER: The only comment that I was going to make, Debra, is my experience has been that I know probably 100 families with rare genetic disorders, and I would say a very small percentage of them have actually got genetic counseling. I think until we resolve the issue of families getting better information from their more general practitioners and getting referred to genetic counselors, I think we need to resolve that issue before we tackle the need for more genetic specialists.

DR. LEONARD: It's just that in anticipating the response, you train the generalist, and the competencies are not listed that they have to know how to do the interpretation of the tests. The competencies generally say you recognize when you need a specialist and you refer. So we're going to train all the generalists to refer, but there's not going to be anybody to refer them to. Or is there? Hunt, you may have a better sense of the numbers, but I'm hearing from the National Society of Genetic Counselors and the American Board of Genetic Counseling.

DR. WILLARD: Part of what you said I don't disagree with. What I think is still a contentious issue is whether the only people who "have sufficient knowledge of genetic testing" will be those who are genetic counselors or certified medical geneticists. I do not think you get buy-in to that conclusion perhaps even around this table, much less if you went very far outside this room.

DR. LEONARD: Well, true. I'm neither of those, and I do genetics. So there are other organizations and groups that do this. So maybe it's not genetic counselors or medical geneticists, but we have not at all addressed -- I mean, this is all general, and if that's what this document is supposed to be, that's fine. But I would like to remind the committee that I don't know if there will be enough specialists as we move forward.

DR. WINN-DEEN: So perhaps what we could do is add one bullet point that just addresses the need to, without saying specifically how large this number needs to be, but in support of this expansion of genetics and genomic medicine, that we also need to train an appropriate number of referral specialists.

DR. REEDE: Sarah?

MS. CARR: Well, I just wanted to ask Suzanne Feetham if you could recall for us what the scope of the workforce analysis is that HRSA is doing now and that we heard about in October, and whether or not it might be prudent to wait for the findings of that analysis before making recommendations about specific specialties. Will that analysis make a determination about whether genetic counselors are going to be in under supply, or are now?

DR. FEETHAM: There were two studies. A study that was completed a couple of

years ago was on genetic counselors, and in that study it did acknowledge that there was a small number, 1,800 at that point in time, and that that was insufficient to meet the needs. But there was also attention, as I recall, in the recommendations that where this was going into primary care, that quadrupling the number was still not the issue. It was what we've been talking about.

The current study is looking at genetics in primary care and with a specialist, and I don't expect that it will be more of a description of a practice than giving numbers. I don't expect numbers to be coming out of that, but I would expect that, again, what we've been hearing is that we will need and always need genetic specialists, and we have a high need for this knowledge in primary care. It will give us more description of the practice by both genetic specialists and primary care in this current environment.

DR. REEDE: Ed?

DR. McCABE: I was going to comment that --

DR. FEETHAM: Did you have another --

MS. CARR: My recollection of the first study was that it didn't draw any conclusions about the adequacy of the supply, and looking to the second study to provide more specific recommendations in that area. But if it's not going to do that, then it's important for the committee to know that, I think.

DR. FEETHAM: Well, I don't think it's the type of study that's going to come out and give you a ratio of specialists and primary care. I mean, it's more a description of the practice base and hopefully giving a baseline for the future.

DR. McCABE: I think we're a long way from Debra's concern that we're going to have so many generalists educated in genetics that we are going to overwhelm the specialists in genetics. So I would either leave it general, which is the tone that we have it here. If we were to insert something, I would say something general like evaluate the adequacy of the specialized genetics workforce. But I'm not sure we need another workforce study. We've had those, it's inadequate, but it's a different issue than inserting genetics education across the board, which is really what I think this is about.

DR. HOOK: And along that line, it's not an either/or. It's not generalist versus medical geneticist. I mean, a lot of the counseling that's going to take place is coming from other subspecialists -- neurologists, hematologists, gastroenterologists -- who you indicate at earlier points should have as part of their training a very thorough understanding of the genetic anomalies within their organ system subgroup, and that's also a referral base that doesn't have to go to a specific medical geneticist but that you have covered in the principles that you've previously articulated.

DR. REEDE: Alan?

DR. GUTTMACHER: No matter where one comes down on the question of specialists versus generalists, I think in the end it actually is more pertinent to the resolution on reimbursement. We've treated that reimbursement largely by genetic testing. I think the real driver of the number of genetics professionals is not the training programs. People are in training programs for a couple of years. You're then out practicing for a lifetime. It's who gets reimbursed to what degree for doing what that will actually drive the need for genetic counselors and medical geneticists, primary care people doing genetics, et cetera. That's much more the driver, I think.

I think that no matter where we come out on this, if we're really going to influence that, I think it's probably more important to influence on the reimbursement end rather than from the educational funding end.

DR. REEDE: Any other comments or suggestions with regard to the recommendation?

(No response.)

DR. McCABE: So could I take it, then, that we will leave the specialized genetics

training out of this, that we'll focus more on improving genetics education and training across the board, and that that will be another issue for a later day? Okay.

Thank you very much, Joan.

At the last meeting we talked about large population studies and pharmacogenomics. These were identified as needing in-depth study. I want to now talk about a meeting. There was a March meeting. At our March meeting, Francis Collins invited us to appoint a liaison to the NHGRI working group organized to consider large population studies. Chris Hook was appointed as our liaison to the NHGRI working group, and I'd like to thank Chris for taking on this responsibility and ask Chris to update our committee with respect to the working group's activities, and then have Alan Guttmacher following Chris update us on efforts by NIH on this issue.

Chris?

DR. HOOK: Thank you, Ed. I'll be brief given the number of things you still have to cover.

The working group has had two face to face meetings and one phone conference to date, and I want to thank my colleagues in the AGES group for setting up a phone line for me because it's been very difficult to travel with some home front issues. But the discussions have been lively. It has been an education in population genetics and the incredible logistical concerns that are covered in this. Obviously, it's still a work very much in progress.

Covering the broad front of issues, such as the ability to utilize existing population cohort studies in a new collaborative or consolidated sort of a fashion versus starting a new project on its own with all of the issues of trying to secure funding and so on for a project of that nature. There are very complex issues in terms of understanding the power, understanding recruitment, single versus rolling informed consent, dealing with a population base which essentially could be followed for 50 to 100 years over the lifetimes of individuals, from infancy to their demise, and how do you account for the changes in the technology that will take place during that period of time, and a variety of other significant issues.

There is at least one more face to face meeting planned in August, I believe it is, and some more phone conversations before a report will be rendered by the group.

A couple of points in regard to the handouts that you've received. There is Dr. Collins' article in Nature from May 27, which lays out at least some of the issues that the group is considering in trying to develop this large cohort study, and a modification to the request for information for public comment has been extended to June 30th. So they are still receiving statements from individuals concerning that. They have already received a significant volume to date, but obviously with the number of questions that are on the table, there is still interest in getting further input.

I think at this point I'll turn it over to Alan for further comment.

DR. GUTTMACHER: Thanks, Chris, for your involvement in this, which we have really appreciated.

I'm not sure there's a whole lot to add to Chris' very good summary. The information he mentioned to you is in I think the table folders that were provided this morning. Again, I would just, as Chris did, call your attention to the request for information, which is open for another couple of weeks. It has 14 specific points. It requests folks in the scientific community and the public who have any expertise or points they'd like to make about these 14 questions specifically or other questions they think would be of use. We invite comments. There have been scores of comments received already, and they've actually been very helpful to the working group to see the breadth of this.

A lot of this does get to the question of how one works with existing cohorts because there's certainly a role for those, but also to think what are the ways that a new cohort might add to our understanding of all this.

I don't think there's really a whole lot else to add. We hope that by the end of the

summer/early fall, this working group will have achieved its purpose of trying to really figure out the science and to a small degree the logistics of what such a study might look like, and then it will be up to higher-ups at the NIH and other agencies of DHHS and the administration to figure out whether it makes sense to go forward with such a study or not.

DR. McCABE: Any questions for Chris and Alan about this process and the large cohort study?

(No response.)

DR. McCABE: Please take a look at those 14 points, and if you wish to have input to them, please respond to them.

Okay, thank you.

The next thing I'd like to talk about is the agenda for the next meeting, and then we'll talk about other issues beyond that. Yesterday we talked about including on the agenda at the next meeting two processes or two groups of individuals, and I have them down as genetic discrimination, bringing real people with real problems related to genetic discrimination. That was the terminology that was used, and we specified some specific types of genetic discrimination, and we talked about speaking with the Genetic Alliance and other groups to help us identify those individuals; also talking to Paul about if he could help us identify individuals that have come through the EEOC.

The other group that we had talked about like that were coverage and reimbursement, again identifying individuals who -- I have it down as the impact on the health care for individuals refused coverage and/or not reimbursed for genetic testing, genetic services. So this would also require some identification of those individuals, but we had talked about both of those groups yesterday.

Then the options that have come up in our deliberations today are an update -- again, these are options, and we can't fit them all into the agenda, so we'll have to make some selections -- an update on the CDC working group effort for ACE+, the process of establishing CPT code reimbursement, both of which will relate to the draft document that we're hoping to finalize at the next meeting; update of the National Academy of Sciences group on patent and licensure, and then large population studies, if that had been finalized.

So what is everybody's wishes? I think if we do the genetic discrimination coverage and reimbursement, we bring in people who have been impacted by those issues, that's probably pretty close to two half days right there. So we're left with another half day, or we can fill in parts of those half days. They may not be full half days on those, but they're going to be significant investments in time for those.

The first two topics under the options relate to the coverage and reimbursement, CDC working group effort, ACE+, process of establishing CPT code reimbursement. One could argue that the patent and licensure through the National Academy of Sciences is relevant to that. Large population studies will be completed by mid-October? If it's up in the air, my guess is it probably won't. So maybe we could postpone that to the following meeting and just put a placeholder in the following meeting for the large population studies.

So now we're down to three.

MS. CARR: I think we also have to build in time for a review of the revised version of the coverage and reimbursement report. Is it the committee's hope that we will revise the draft and go out for public comments with the revised draft, or do we feel we need to sort of work on it some more and then look at it again in October and then go out for public comment?

MS. BERRY: The latter.

MS. CARR: The latter. Okay.

DR. McCABE: Any disagreement on that? The head of the task force said the latter.

MS. BERRY: The task force of one.

MS. CARR: And maybe that should be broadened out.

MS. BERRY: I think that would be a good idea, because then we can get everyone's input, and that might make our review more efficient so that we could have a broader group, everybody look at the revised version once staff has had a chance to put that together. So I don't know who would want to. Are you going to force people to join?

DR. McCABE: Do we have volunteers? If not, we will force people to join.

Put Debra's name down.

DR. LEONARD: I want to ask a question. Since the committee has gone over this document, is it something that the whole document, once its revised, could go back out to the entire committee and ask for comments back? So that the discussion time -- I mean, basically doing the discussion by email, electronic communications. I don't know if that works for everyone or not.

DR. McCABE: I think Cindy was hoping for some additional help in reviewing the next draft to make some changes there before it went out to the committee. I think given the amount of work that staff has, this is unlikely to happen within the next two to four weeks, in which case it's going to be coming back fairly close to the time of the next meeting.

Do I have any volunteers in addition to Debra who wish to?

DR. LEONARD: I've already been put on one task force, and because I might be --

DR. McCABE: I'm just teasing you, Debra.

Anyone?

DR. WINN-DEEN: I'm willing to volunteer, and specifically I think we need a few people who can help if staff, as they try and translate all our discussion here into things, has some questions they need to just bounce back and forth, we need a little referral group to just help them with that.

DR. LEONARD: Well, I did already volunteer to help with one part of it, which I will do.

DR. McCABE: Okay.

Anyone among the ex officios to help with that?

MS. CARR: Maybe Muin, since we're going to try to incorporate your -- and I think we should volunteer Reed Tuckson in his absence.

(Laughter.)

DR. McCABE: In keeping with the tradition of all committees. Reed had a lot of input, too. So it would be good to make sure he's satisfied with how his edits are incorporated. That's how we'll justify doing this to him.

So that gives us three topics that are clear, genetic discrimination, coverage and reimbursement with individuals, as well as the review of the revised document. CDC working group effort, ACE+ -- how long will it take you to do that, Muin?

DR. KHOURY: It depends when the committee wants to hear. October will be at the beginning of this next phase. If you want an abbreviated version, maybe we can take a half hour or less. If you want something more of a discussion, I think it will be an hour-plus. But I wouldn't think you'd need more than half an hour.

DR. McCABE: Okay. If you can do it in a half hour, that would be great. So we'll allot 30 minutes to that.

The process of establishing CPT code reimbursement. Are people interested in that for the next meeting for our education?

DR. REEDE: I think if we're going to be talking about reimbursement, that it would be nice to understand that better.

DR. McCABE: Okay. So we'll try and identify someone who can speak to that

process.

DR. LEONARD: I would suggest Mark Synovec because he does that, and that's practically his professional life's work.

DR. McCABE: Can you provide his contact information to Sarah and her staff, please?

DR. WILLARD: Can we clarify? He's a pathologist who is trying to fight that system, or is he on the inside of that system trying to help?

DR. LEONARD: He is a pathologist, but he works with the AMA CPT Editorial Board, he works with the reimbursement side. He has gone through this process many times and understands how it works. At least we could contact him. If he's not the one to speak, he could say who in different organizations, government agencies or whatever, are the appropriate people to inform us.

DR. McCABE: And likewise, I know there's somebody -- that would be from the pathology side, and then the American College of Medical Genetics has someone from the medical genetics side, and I'm not sure who that is.

DR. WILLARD: The reason I raised the point now, and I would bring it up also in the context of ABMG, is that to me it actually would be useful to have someone on the other side of the desk who can explain why it isn't just as simple as saying this is what it actually costs, and gee, I'd like to be reimbursed for 100 percent of my expenses, because I suspect people on the other side of the desk will just shake their head and go where are you coming from? This doesn't happen. So it has to be someone on the other side who sits there and does the thumbs up and thumbs down on these things.

DR. LEONARD: And does it across the board, not just laboratory tests but genetic services, genetic counseling, all of the aspects that are related to genetics.

DR. McCABE: Okay. Then we have two topics. One is in preparation for February. As people are leaving, we can volunteer them now for these other work groups.

(Laughter.)

DR. McCABE: If we're going to talk about pharmacogenomics, then we need to put together a group to address the topic of pharmacogenomics. Anyone who wishes to volunteer for this? So Emily with that group. Anyone else? Anyone who is left who would like to?

DR. WILLARD: It's not clear to me what you're asking. You're not looking for speakers.

DR. McCABE: This is to plan for --

DR. WILLARD: Plan that session.

DR. McCABE: This is to plan a session on pharmacogenomics.

DR. WILLARD: I'm happy to do that.

DR. McCABE: So we're talking about Emily, Hunt, Debra, Chris. That's the group. And Suzanne. Anybody else among the ex officios who I may have missed? Kay.

So we're left with the one topic we haven't decided.

Oh, Sarah is telling me we need a group for large population studies. Who would like to do that? Chris, I think you're stuck with that since you are our liaison. Any interest in that?

DR. WILLARD: Interest, yes, but do I want to be on both of them?

DR. McCABE: Can we call upon you, Alan, to do that, and Muin?

DR. LEONARD: For pharmacogenomics, should there be someone from FDA on that work group? That would be useful to inform us.

DR. WINN-DEEN: There could be. I just didn't want to volunteer Steve.

DR. LEONARD: You can always volunteer Steve. He's not here.

(Laughter.)

DR. WINN-DEEN: There's a lot of FDA guidance documents that are going to be

coming out with updates. Maybe you want to say something about what the timing of those are so that we can plan an update to this committee as well.

DR. LEONARD: I think just putting somebody from the FDA on this work group to help plan the pharmacogenomics sessions for February would do that. I don't think we have to discuss it now necessarily.

DR. WINN-DEEN: There's more than just the pharmacogenetics guidance document that's going to come out. Microarray and companion diagnostics are also on the list, right?

DR. MANSFIELD: Did you want to know that now?

DR. WINN-DEEN: Would you be prepared to do something in October, or should we just plan for February?

DR. McCABE: I don't think we have room in October, unfortunately.

DR. MANSFIELD: The voluntary genomics submission is supposed to be finalized this year. It's sort of creeping. I don't know that anything else will be finalized this year. The combination therapeutic diagnostic workshop is going to be in July, so maybe a draft will be out this year.

DR. McCABE: Okay, good. So you just volunteered Steve, I take it. Okay. Let the minutes reflect that.

Then Sarah and I just conferred and we think we could do something very brief on the National Academy of Sciences' process on patent and licensure. We could fit that in in October also. Anybody who doesn't want that in there?

(No response.)

DR. McCABE: I think it would help with the coverage and reimbursement, actually, because it is part of that story. So something very brief, probably another 20 to 30-minute process.

Are there other topics that people would like to bring to the floor? Other topics that we should be thinking about with even longer-range planning?

(No response.)

DR. McCABE: If not, I think that wraps us up. Everyone travel safely who is traveling, either short or long distances from here, and we'll see you in October. Thank you.

(Whereupon, at 2:39 p.m., the meeting was adjourned.)