Questions and Answers with Edward McCabe and Sarah Carr

DR. McCABE: Questions for Sarah?

DR. BEMENT: I have an observation. As I looked at this list of top 10 issues, there seems to be a commonality in at least seven of the issues, Issues 1 through 4 and 8 through 10, in that they bear on the public trust and confidence, not so much concerning where it is at the present time but the extent to which it could be a barrier or the means necessary to build public trust and confidence in order to move ahead, and so that crosscut might be worth also focusing on.

DR. McCABE: Yes, and that's one of the things we were going to talk about, is it's important for us to begin to identify those intersections because some of these do flow together. For instance, the one that was identified by eight agencies, is quite broad and that could be part of why it was identified by so many of the agencies.

But it's important, as you see these groupings, natural groupings, please help bring those to our attention because this is important for us to look at and may help us in identifying priority areas because we're not obligated to follow these specific areas. For instance, if we took that one that was identified by eight agencies, we'd probably have to focus on more than that because it itself would be too broad. So to the extent that we can redefine some of these categories in order to focus, that will be a valuable exercise for us.

DR. TUCKSON: For the record, I would like to indicate that on the slides, under a number of specific issues identified within seven functional categories, the next-to-last bullet, Use of Genetics in Bioterrorism, is not meant to be literal nor in Slide 9, Point 9, on the other one. So I just want to make sure. You never know who will read these things in the public record and decide that what we're here talking about is use of genetics in bioterrorism. It is a minor point, but knowing the way the world works today.

DR. McCABE: Thank you.

Other comments? Ideas of the Committee members or the ex officios on how we proceed to basically slice and dice to get to the priority issues? Reed?

DR. TUCKSON: I don't know how to get at it. Maybe it's in the material for our discussion today, is I think one of the questions that you asked, Sarah, and that Lana did, was what the relationship is between what's already being well addressed by the agencies, and I don't know how to tell from this summation -- can we assume that if it's on this list, that it says that this is an area that the agencies themselves feel like is not being addressed? Can we imply that?

Secondly, we need to be somewhat clear about which of these -- we are an advisory Committee to the Secretary of Health, and knowing that they all talk to each other, but our only portfolio is what can the Secretary of Health influence, I assume, or cause to be influenced, and so one of the things that's sort of a sense is that how much of that do we need to be thinking about in terms of looking at this list as, i.e., the agencies are saying we need more attention to this, therefore Secretary of Health, please, you need to be helped in getting this done.

MS. CARR: Well, your charter actually -- and there are 16 agencies, nine of whom are in the HHS but there are seven that are in other agencies. So the charter says that you're to advise the Secretary of Health and other agencies on request. So if there are issues in another realm that you think are important, it would be important to understand whether that department would be receptive to your advice, but I guess I don't want us to think that you can only speak to the Secretary, and I think copies of your reports and recommendations will be taken back by the other ex officios to their agencies because they may pertain to a health arena that relates to our Department but they may have some application, at least relevance, to the other agencies as well.

MR. MARGUS: So I also wonder about what's already being addressed, and then I realize also that people at these agencies are smart people who think about this all the time. So what are we supposed to add as a Committee, and I guess they have their fingers and tentacles out in the whole country. So it would be arrogant to say maybe we can bring our outside-of-the-Beltway perspective but maybe there is something else we could do, and one was maybe thinking looking forward, are there any trends coming down the pike that we may be able to anticipate now rather than just thinking about what it is right now. So I'm not saying I can foresee those but maybe we should at least do a little ideating about what else could be coming that might affect it.

A couple of things we've heard about on the science side, we're going to shift now toward complex common diseases that are likely to be caused by multiple genes, either working in an additive way or a combinatorial way, and so genetic tests are going to get more complicated and then the utility of those tests might be a little more complicated, too, in the sense that you may not have as much of a certainty in the results, that there might still be use in them. The science would be one area to consider.

Another is what everybody eagerly bashed yesterday, which was the website things, but I didn't really hear the answer on who oversees marketing of genetic tests, but separate from all what seems like, as a layman, really rigorous review of clinical tests and how they're done and all that, then it feels like there's a lack of review on how they're marketed, and I know we don't like the bad science, junk science stuff, but there's no reason that there can't be other channels for it, but that's something kind of futuristic. The day may come sooner than we think when we'll have to care about that, and I don't know who's in charge of all the fine print that's mentioned when you see an ad for a drug company's product on TV these days. After the people run through the field, you have 50 sentences about all the side effects, but who's in charge of doing that for web marketing and everything else for genetic tests?

But those are just two areas, the science base or maybe on the consumer marketing base, but I think it would be a good exercise to go through just to think about anything else that's coming and maybe this Committee could come up with a statement on it, maybe not, but it wouldn't hurt for us to make a little effort in identifying those things.

DR. McCABE: I think certainly the direct-to-consumer marketing is complex and we did have discussions, but I think all that really showed me was how complex it was. Other than the fact that the regulation is fragmented, we didn't really get into that, but that's certainly something, given the audacity of some of these claims that we saw, that's certainly something we could explore, and so we can put that on the table as one of the things to look at as we move forward.

DR. WINN-DEEN: So I guess from my point of view in terms of time well spent, I would really like to see this Committee be focused on working on issues where we can make a difference and not just serving as a two-day discussion of stuff. Either it's not in our purview or there's really, no matter how much we talk about it, not too much to be done. So sort of in the category of things where we could make a difference or maybe some of the things like the genetic counseling group brought up this morning where

there's some specific recommendations that they may come back to us with, which we can then debate and determine whether we as a Committee feel we want to make some very specific recommendations about how to advance the education of health professionals and the recognition of genetic counselors as health professionals.

I don't mean that as a single agenda item but that's the kind of thing where I think we could do something very concrete. We could have a Committee discussion and we could move ahead.

There are other areas, and again I'll just pick an example, where I think the issue of what the U.S. Patent Office deems to be patentable is probably outside of the charter of HHS or this Committee, and while there should be debate about that, I'm not sure that that's the kind of thing that we should be focusing our time and energy on. I think that the aspect of that that has been highlighted, which is, is there a barrier to access, that's where we should say that there's an issue there on access and health care provision to all, but I don't want to see us get sidetracked on things where we just are not going to be able to make any real progress and to have this Committee at the end look back and say what did we really do with all that time we spent together?

DR. FELIX-AARON: I was reviewing the third to the last and the second to the last slide, and it was not apparent to me the connection between those two slides. So for example, the first bullet in the third to the last slide says integration into health care and public health care systems. I mean, that got 23 hits, but when I looked on the top 10 specific issues, none of those issues seemed to relate to the integration to care issue.

I make that point because a number of the things that have come up today focus and are directly related to the integration into health care. So the workforce issue, sort of the translation and implementation issue and access issue are clearly issues that yesterday and early today were brought up, and so I was just wondering how were we going to integrate sort of what we learned today and sort of the important themes that emerged into this priority list.

MS. CARR: I think you're identifying probably a part of the problem with summarizing that data in the way we did, but we also looked at it in another way, which was by the functional category, and when we did it that way, for example, seven agencies identified the integration into health care and public health. That's where most of their issues, if you look at them as a whole, fell, and so from the standpoint of the functional categories, that did seem to be the one that most agencies selected, and the seven were primarily the HHS agencies, but also DOD.

So I don't know if that helps at all, but I think you're pointing out something that we just cut it one way and there's a lot of ways to look at it, probably that data, or maybe not. Maybe we overinterpreted or beat it to death.

DR. FELIX-AARON: Right. I wasn't criticizing the methodology.

MS. CARR: Right. I know.

DR. FELIX-AARON: I was just trying to make the connection between sort of what is here and what we learned today and how what we learned today will help us in terms of determining sort of the two types of analyses and the way to move forward.

MS. CARR: Right. I think I'm revealing probably more of my misgivings with summarizing it.

DR. McCABE: So in follow up to what Kay said, what have we heard in the past day and a half that fits along with these priorities or other priorities that one might want to establish? I'll just throw one out there and that was from Kim Monk this morning. We heard the status of the genetic nondiscrimination. That's the kind of thing that if the Committee so desired, I think with Secretary Thompson's support of that legislation that we have here, dated in May, as well as prior indication of support that he gave us, it would be appropriate for this Committee to thank him for following through with that support and discussing what we understand to be the progress and offering our support for any future efforts in that area, but that's the kind of thing that we could do, based on what we've heard, but I'd like more discussion from you.

DR. LEONARD: Well, I agree with Emily completely, that we should focus on issues where we can have an impact, and the other point is when you look at something like integration into health care and public health, what are the problems? Are we anticipating that there's going to be an issue or have problems actually been identified that need to be addressed?

So I think if we focus on areas where there are issues, where we can have an impact, it would be better, and I don't know how much time we have, but I made a list integrating everything over yesterday and the first was discrimination protections at the national level, and I agree that we should send a letter from this Committee. I agree with you that we should support moving through the vote in the Senate and then moving it through the House so we can actually have a law in place. Like Emily said, until it's law, it doesn't do anybody much good.

The second issue is oversight of genetic testing. Since that was such a major issue with the SACGT, I don't know. There are recommendations that are in place that may or may not be being acted on. I don't feel that we got a lot of input from the FDA on where they're progressing with theirs or with CLIA, and so if FDA could be included in the October update, I think that that would be useful as to what they are doing. They seem to be focusing on ASRs rather than coming into laboratories, but from discussions here on oversight of genetic testing, there seems to be more concern not on the CLIA laboratories and the analytical testing that's being done but more on the postanalytical interpretation on the ordering of the tests.

So I don't know how everyone else feels, but with CLIA revising their regulations, I know I saw the draft of the original changes that they were proposing to include for genetic testing, and it extended beyond what the laboratory can do. So I don't know if this Committee can recommend to CLIA in their deliberations and in their new regulations to really focus on the laboratory rather than moving the laboratory as an intervenor into the doctor-patient relationship, which is very hard for laboratories to do, and then your suggestion that we focus on truth in labeling on genetic testing rather than actual FDA regulation.

DR. McCABE: Steve Gutman is here representing FDA for David right now.

DR. LEONARD: I know.

DR. McCABE: And was very involved with SACGT on the recommendations from SACGT. So perhaps you can comment, Steve.

DR. GUTMAN: I'd like to offer that the FDA certainly would be willing to come back at the next meeting and provide a more comprehensive briefing, and in view of the fact that some of the members are new and perhaps not familiar with our process, provide some background for what we do. We probably

think it would be appropriate to share the stage with the folks from CLIA so that they can actually also clarify what they do, and I don't know where it'll be in terms of their revisit of their activity, but it might be highly timely.

We also might suggest, if you're really trying to look at the comprehensive selection of the options to get hold of this, it may be FTC might be brought in as well, so that there could be information on -- FDA does, particularly once it's classified something as a device, have some authority over the promotion, but obviously that's a major FTC effort, so they may be a player as well.

From our perspective, when the previous Committee, SACGT, the total product life cycle ran out and it was recycled into this new one, the Committee disappeared. The recommendations didn't, and as it's been publicly acknowledged, and I'll certainly publicly acknowledge it, that our agency is very hard at work on trying to move forward with the spirit of the recommendations. It's easier said than done. We've gotten queries from a variety of people, both in the manufacturing community and in the professional labs with notions that they'd like to leverage or partner, and we take those opportunities seriously and certainly input from this Committee on trying to provide us your insights in the direction we're going and how you view it would probably be quite welcome.

DR. WINN-DEEN: Steve, could we potentially also hear from somebody like Larry Lesko about the pharmacogenetics side?

DR. GUTMAN: Absolutely. Of course.

DR. McCABE: Just so we can really formalize this for our record, the people who we would want to invite would be FDA, CLIAC, FTC, and FTC is not represented among the ex officios but we can approach them, and perhaps among the ex officios we would know who would be the right person to approach in FTC. I don't know about that process, whether we'd have to go through the Secretary's office. There's probably a protocol that we have to do when someone isn't represented here and we can explore that protocol, and then CLIA's responsibilities are shared between CDC and CMS, is that right, and certainly it's important to have their perspectives also.

DR. GUTMAN: It would really be important to have Judy Yost here, actually.

DR. McCABE: I'm sorry?

DR. GUTMAN: It would be very important to have Judy Yost here.

DR. McCABE: Yes, from CMS, Judy Yost. So that gives us a group of five.

I would suggest that we really focus on the status of regulation and regulatory progress at that meeting. I think we want to be cautious not to -- I was tempted to let's throw in the rare diseases and maybe we should get into the pharmacogenomics and those sorts of things, but I think we should focus on sort of where we are on the regulatory process because if we get too diffuse, I'm afraid we may miss where we are.

I think your suggestion, Steve, to give us a primer on the process -- because I know I get lost with some of the acronyms and everything. So I think that would be helpful, too, if you or one of your colleagues would be willing to do that.

DR. BEMENT: Trying to tie those two threads together and integrating health care with public health, one of the integrating elements, of course, is data and information exchange, and this gets into data formatting standards, privacy, encryption, networking, and I presume that would be part of the standards and regulatory discussion presumably.

DR. McCABE: I had some others listed here, but I think I want to follow up on this and then I'll come back to the others who might have taken us in a different direction, but let me just check.

Comments on this topic that we're discussing now? Agnes, was it a new topic or was it on this topic?

MS. MASNY: Somewhat on the same and somewhat different.

DR. McCABE: Well, why don't you go ahead?

MS. MASNY: Well, my first comment was that I wanted to just agree with your point about supporting the legislation and writing to Secretary Tommy Thompson to thank him for supporting the bill and that that would be something very specific we could do because I think that it came up by many speakers, the issue of genetic protections against discrimination as well as for privacy, and just to sort of reiterate what other speakers have said and from my perspective in my own clinical practice, that it is true that because of the fear that many patients have that they're not making use of the genetic tests or that fear is still there, so I think that would be a real priority area and something very specific and practical that we could do.

I just wanted to mention, as people sort of start talking about getting into specifics, is that because I think that the work that we were given to do and the charter that we were given to do looking at how genetics will impact health and society, that I think that maybe we should look at ways that we could also have sort of a broad focus as well.

One of the things that I was thinking of as we heard the speakers yesterday and today was looking at ways that we could actually see genetics be more integrated into health care. I mean, that's one of the priority issues, but I'm thinking that there was a statement from the Task Force on Genetics and Insurance that was a subcommittee of the National Human Genome Research Project ELSI Group stating that if we looked at the genetic information as different from other medical data or other health information, it's conceptually confusing, practically infeasible, and ethically indefensible.

So I think that as we're approaching some of these things, if we look at genetic information totally in an exclusionary way and not integrative into how genetics will impact all of primary care and all of health, that I think we need to move in some way to help both the public and, as we mentioned about the workforce issues, of how genetics will be integrated and that we have to think of it as just another tool for medical and health care practice.

I talked yesterday evening at our dinner meeting to Mr. Tim Baker from the CDC, and I think one of the ways that that is already being done with the CDC is their way of incorporating and integrating genetic information into most of the bulletins that go out. So they now have a special section on how that's being integrated into all the CDC's information.

So that would be one way, and then as we integrate information, genetic information into health care and public health practice, I think that the issue of the validity of the interpretation of test is going to be critical. So that, if we have that briefing and we have more updates on the clinical utility and where we are with that, maybe the one thing that we could focus on would be as some of the criteria or a way to look at evaluating the validity of the interpretation of some of the information that we're going to be

getting not only from single gene testing but the multiplex testing and the assays that will emerge from tumor or tissue in the future.

DR. McCABE: You know, we were headed down the path in terms of what we were going to do the next time. Debra, now you have affirmed the proposal that we basically send a thank you to Secretary Thompson.

DR. LEONARD: Well, not just thank you, but actually somehow supporting this further -- I mean, thanking him.

DR. McCABE: Right.

DR. LEONARD: But also, if there's anything that can be done to facilitate the process to law.

DR. McCABE: Okay. So could I have that as a motion, so that this would be something we could move forward?

DR. LEONARD: I so move.

DR. McCABE: And I'll take your comment as a second, Agnes.

Any further discussion on this? Clarification?

(No response.)

DR. McCABE: Given the need to move forward, if this is going to be dealt with after the July 4th recess, then it would certainly be something we'd need to move forward before our next meeting. So if we have a sense of what would be included in that, and if you could give us the go-ahead, meaning Sarah and her staff working with me, to move that forward. Would that be acceptable?

DR. LEONARD: Yes, and can the letter be sent out by email, so we can look at comments?

DR. McCABE: Yes, and we'll certainly have it out.

DR. LEONARD: But it should definitely go before our next meeting.
DR. McCABE: Yes.

DR. LEONARD: And in fact, as soon as possible.

DR. McCABE: Any further discussion of that letter?

DR. WINN-DEEN: Who's going to write it?

DR. McCABE: Who's going to write it? Staff will write it for my signature. But we will have it reviewed by the Committee.

MS. CARR: But if you would like to, you're welcome to.

(Laughter.)

MS. CARR: Yes, it will be signed and it will come out to you all for review before it's finalized.

DR. McCABE: And I could tell you the staff is extremely articulate, and then with the input that all of you will have to refine that.

Any further discussion?

MR. MARGUS: When you invite these people to the next meeting, besides this --

DR. McCABE: Why don't we deal with --

MR. MARGUS: All I want to say is we give them a primer on what they do. It would be great if they came just with some thoughts on what they thought was broken.

DR. McCABE: Okay. Motion we have on the floor. Any further discussion?

(No response.)

DR. McCABE: All in favor, say aye.

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Abstain?

(No response.)

DR. McCABE: So we will move forward on that letter to Secretary Thompson. Thank you very much.

So now, going back to the next meeting, the other group that -- I don't know if it should be in there, but if we're getting into the advertising side and with FTC, does Commerce then play a role?

MS. CARR: Well, FTC is an independent agency. So are you thinking other agencies within Commerce that would also be involved?

DR. McCABE: No, no, that was anyone.
MS. CARR: Anybody?

DR. BEMENT: I don't know. We could perhaps help with a workshop to get input from the private sector. It seems that a more appropriate role would be to look at it from a technology policy point of view.

DR. McCABE: Okay. Good.

MR. MILLER: Just to go in a little bit of a different direction in thinking about what this Committee might sort of intersect with an issue that's been raised a number of times, both this morning by Ms. Monk and throughout the day, dealing with the issue of genetic information and discrimination and insurance

and employment and also added on here education and law, a number of people, both on the Committee and others, commented that there's really a dearth of information, of experiential information. There's not a whole lot of sort of cases coming forward complaining of discrimination and everybody is sort of foreseeing this as a problem sort of around the corner.

I wonder whether it might be an appropriate use of this time to sort of get to use the Committee and the various different points of view from the Committee to really get a sense of what is the concern out there, how deep does that concern go, how many people do feel that they're currently experiencing discrimination, to get a larger sense for the scope of the problem as it exists today because there's not a lot of information out there about that.

DR. McCABE: Now, these data are old. They're probably three plus years old and they were anecdotal.

MR. MILLER: Right.

DR. McCABE: But when we approached the public, this was a major concern, and as has been mentioned, while the number of cases are relatively small and tended to be among the self-insurance where the employer is also the insurer and you're certainly aware of that since you've been involved in those.

MR. MILLER: Right.

DR. McCABE: The public has extreme concern and is having the testing done anonymously or under pseudonyms because of their concerns and that creates certain problems as well.

This would be the kind of thing that we could certainly explore, though I wonder if perhaps you and your colleagues have already explored this and written about the cases that have come before the EEOC.

MR. MILLER: The issue is that there really haven't been any, I mean, with the exception of one case which got a lot of notoriety and which we prosecuted and resolved against a railroad. There really haven't been any cases that have been coming forward either to the EEOC complaining of employment discrimination on this basis or, quite frankly, in any of the state agencies, the state human rights commissions, that enforce any of these state statutes.

There has been anecdotal evidence and stories and some of the consumer groups talk about it and there's a lot of anecdotal talk about the existence of discrimination, but there isn't really a lot of cases that have been coming forward in any sense and there's a great deal of confusion and question about why that is.

One of the issues that has been sort of chewed through or put on the table in terms of some of the public discussions around the legislation and these other issues around discrimination is that it's really premature in that there are really very few cases coming forward. It may be worthwhile to go back and to explore some of those issues.

DR. McCABE: There was another one, which were the alpha-1-antitrypsin cases, though that one was under the ADA.

MR. MILLER: As was the railroad case, but the case with the alpha-1-antitrypsin never went forward into a lawsuit.

DR. McCABE: One of the things, also, that I've learned from you is the constraints on time on the reporting.

MR. MILLER: Yes.

DR. McCABE: And it might be good at some point -- Sarah was commenting that the Alliance has been exploring this, and it might be good to --

MR. MILLER: Or even to explore barriers, to the extent that there are anecdotal stories of evidence of discrimination that have occurred out there that people talk about and that groups know about but that they're not being translated into either enforcement of rights or what have you. If there's a sense of barriers out there or if there's a sense that people just don't believe that there are any laws currently protecting them or there's a lack of knowledge about sort of what their rights are or if there are other issues in play to try to better understand the issue of discrimination in this area as it appears that the sort of legislative train with respect to discrimination bill, whatever it may be, I mean, that's occurring on a separate track and there is little that this Committee can, I think, do to get involved in that process.

DR. McCABE: So that, perhaps we could have staff confer with the Alliance and with you to look at that.

MR. MILLER: Or the genetic counselor group or some of the other groups that are represented by your Committee.

DR. McCABE: Thank you.

DR. ZURAWSKI: Can I put a little context on that?

DR. McCABE: This is Paul Zurawski from Labor.

DR. ZURAWSKI: For the group health plan environment, we at Labor had taken an audit based on 2001 information of health plans, trying to examine whether they were violating preexisting conditions or HIPAA, including some health status terms, and the truth is that this was not based on complaints or cases. This was a statistically valid random sample type of audit, and there was very few, although, I mean, measurable, but under 1 percent type of evidence of health plans having terms within their plans that would violate HIPAA.

Now, the rules were not yet final and so it isn't in terms of a real-time subject matter. It would be interesting for this Committee to include the health insurance type of aspects of discrimination and we could provide, at least from the group health plan context -- the HHS has the individual market jurisdiction -- our findings, if required or asked for, regarding what we've seen and we have provided some of that information to the HELP Committee as they were deliberating because there was some question about whether or not we, in terms of -- I think last year, we had 184,000 consumer inquiries about their health plan and having almost none of those register as having a genetic information type of concern.

DR. McCABE: Others have had their hands up and these issues may have already been addressed, but let me run through them.

DR. HOOK: Since you're inquiring as to agenda items for the next meeting, I wanted to bring us back because I'm going to focus on the first three points of our seven enumerated functions and just to

articulate support for speaking again with Ms. Bennett in terms of the counselor shortage and seeing if we can move along the process for encouraging the development of manpower. I think that would hopefully be able to be moved relatively expeditiously.

DR. McCABE: And we have asked for information within the next month, so that will be able to help guide us once we have that.

DR. HOOK: Yes. And in terms of our ELSI mandate, I think it would be very, very helpful as we are wanting to or are charged to explore some of these questions and PGD was one that came up frequently in the list from the supporting ex officio agencies, that if we can't fit it in in October, at least in the near future, we should have formal presentations from the President's Bioethics Commission and from the HGP ELSI Project on the status of where they are on some of the issues that they are discussing of this nature, such as the difference between treatment and enhancement and trait selection and things of that nature, so we're not reinventing the wheel but knowing where our colleague organizations are in their study of these issues.

DR. McCABE: Thank you.

DR. FELIX-AARON: I would also like us to consider in terms of the issues that we focus on a balance between macro policy issues and practice level issues, because I think a number of practice level issues, issues that patients and providers come up against every day have emerged here, and just to point out a couple of them, the issue of workforce development, the issue of providers having genetic competence and the number of providers, counseling providers available are some issues that came up.

The other thing that I heard in the area of practice is sort of the guidance that the field of patients and providers need in terms of oral translation from development, transitioning from the development of technologies to the application of technologies and what this group could offer as guidance to that whole area.

DR. McCABE: Thank you.

MR. BAKER: Tim Baker from CDC.

A couple of points were discussed earlier. The complexity of oversight of genetic testing that was well debated, richly debated in the previous Committees, include a couple dimensions, and Reed was addressing it earlier. There was a very complex but thoughtful report about the various elements of that.

So I want to bring forward that a consideration for it, if not at the next meeting, an additional meeting, would be the distinction between the regulatory to nonregulatory solutions for some of these questions. There is clearly a huge focus, and I heard you clearly speak to the emphasis on the regulatory process at the next meeting which is appropriate. The prior discussions have been this acknowledgement that there's a wide array of data collection challenges ahead that can be done through nonregulatory means as well. We have taken some steps forward in working with groups to try to characterize the data that is available through some disease-specific and test-specific considerations of what exists that would characterize clinical validity or clinical utility and how does this give us a potential for a model framework for collection, and then the question is how do you collect that? How do you work with laboratories and needing a stick or a carrot to do that?

So I've put that back on the table for discussion, and we do have a couple of projects that we're looking at, basically to characterize what is known and what is not known, and we think that's an important building

block before we ever get to this notion of a large cohort study. It's sort of like what are these elements necessary? What's the potential framework, and how might we go about it?
The second point I'd like to put on the table is in response to Mr. Margus' question, and actually your question as well. We've struggled a lot with the notion of what does this integration into mean, and particularly for people whose day job is not genetics. It's like how do you make sense of that? Where's the traction point and where's the lens?

So what we find in working with our challenges in public health broadly and with our colleagues in public health and in the state and local settings is given what you're trying to do to understand and prevent asthma in this country, when and how and where will the developments in asthma fit into that, and what is it going to take to understand that context, so it's not driven so much out of genetics as it's driven from asthma as a broader challenge, and then understanding this science needs to be a tool that fills in gaps in knowledge, becomes an enabler toward that solution. What kind of training's necessary in guidelines?

Actually, Dr. Burke is leading a team within the Center for Genomics and Public Health at the University of Washington to look at asthma as a follow-on to a conference we had. We looked at some discreet diseases that asked the question, given these diseases and the pervasive challenge in this country, does it mean anything yet? If it does, who does it mean something for? And if it does, how do we further fill in the gaps of knowledge?

So that's the kind of lens that we're coming at that may be helpful to your deliberations in the future.

DR. McCABE: Yes, one topic has come up, and I just want to comment on it because I learned this with the SACGT, and that has to do with clinical utility. So there's analytical validity -- can you measure the same thing and get the same answer? -- clinical validity, which is how does the test function in a clinical environment, and then clinical utility, how useful is this information in clinical decisionmaking, and whereas the public in our discussions with them, they're ultimately interested in clinical utility. How helpful is this to them, and certainly with some of the website direct consumer marketing, this is clearly an issue because there'd be questionable clinical validity of doing a pH test on the urine for gender selection.

However, it became very clear from organizations like the AMA and others that they consider clinical utility the practice of medicine and staunchly defend the right of individual physicians to practice medicine independently without restriction and without regulation. So we need to be cautious as we use these terms, whereas it seemed fairly clear that people want to know how good is this in my health decisionmaking. That will lead us into some waters that are going to be very treacherous because there are people with major stakes invested in that distinction. So we just have to recognize that.

MS. ZELLMER: I just wanted to say, just like Emily, that I think, I hope that the ultimate outcome of this Committee is that we can focus on areas where we can do the most good, and while I think that protecting consumers against some of these tests, the ego-genomics test, is certainly a concern. I guess I hope that we don't focus all of our efforts in that area. I do think there are other ways of addressing those issues through state consumer protection statutes, things like that, that may be more effective.

I think if you want to help individuals who are dealing with genetic disorders, I think one huge area that I have personally experienced and my experience with other families dealing with genetic disorders is just lack of information of health care professionals, and I think that maybe that's an area that other agencies or other Committees are addressing, but I think that lack of information is a tremendous problem. I think most families I know get most of their information off of the Internet and rarely get information from their health care provider, and I think that as technology, genetic technology moves forward, I think there

are a lot of health care providers who don't understand certainly the rare diseases but I think as we move forward with the sort of more common diseases where there are going to be genetic and environmental components, I think there are a lot of primary care physicians and even some specialists who don't really understand all of the implications and they don't know how to advise their patients to make informed decisions, and I see this as a big problem and I would hope that we would spend some of our efforts on how we get the information out to health care professionals so that they can advise their patients of the best course of action for treatment.

I also think that informing the public would be very important. I think that would perhaps help some of the consumer protection issues that we're talking about, is if we can better inform the public, I think that maybe they'll make better decisions on whether to buy genetic face cream or not. But I think those are issues that I know that have been sort of uniformly discussed by each speaker as these education issues, and I certainly think that from my viewpoint, those would be the most helpful to the consumer.

DR. McCABE: Thank you.

DR. REEDE: This is a follow-up somewhat on what Kim has been talking about and others have mentioned in terms of this integration, this recurring theme of workforce and education and really understanding that much of what I'm hearing is very much in generalities in terms of there's a need for people or there's a lack of education or there's a lack of preparation without any real clear solid data in terms of what type of education is being provided, be it in medical education or continuing education. What are accrediting bodies or boards thinking about doing? What are certifying bodies thinking about doing? What is the status of this now, and what is planned for the future?

As I think or as I look at the top 10 areas that Sarah identified, much of what is included in there are topics that are going to end up being related to the future of education, and how is that being handled now? I think not for the October meeting but I think for a future meeting really being able to deal with the workforce education issues, the status, the need, getting a handle or an understanding of suggestions for what could or should be done in direction would be an important undertaking for the Committee.

DR. McCABE: Thank you.

DR. TUCKSON: I think where we are in this is I think we're just starting this discussion on prioritization and trying to figure out how we will lay that out so we'll know what things we want at the next meeting to start drilling down.

As I listen, I just want to keep also, and as people listen, as we listen to each other and learn from each other in this part of the discussion, there's one area that I just want to also keep in front of us and that is, genetics inevitably will not be something different from health care. It is health care. It has implications but so do so many other parts of health care today for all of the issues that are on our plate.

What I think here is important is to also keep in mind, is that, how we make decisions around the use of this information, the access to it, the affordability of it, and the integration in terms of how decisions are made or not made, I think, is exceedingly important, and I want to just keep the reality in front of us that affects all of the points that we're making, which is still that people can't afford what we have now. We've got 41 million uninsured people now. We've got lots of a context that shapes this new movement going forward, and I urge us to at least keep those in mind as we think about what our priorities are here as well.

I think that that sort of gets me towards beginning to think very carefully about the importance, as Kim has indicated, of how we educate the public to make appropriate choices and how the public can participate intelligently in the clinical decisions that are increasingly complex with their health care team and there are an array then of implications that result from that which we might be able to talk more about.

DR. McCABE: Thank you.

DR. FROHBOESE: Actually, I'm following up on Reed's comments and looking at the Committee's desire to identify both concrete issues as well as concrete ways in which to have a positive impact. I agree that the Committee taking a position on issues, such as writing the Secretary, is very important and will have an impact, but one other thing that I'd urge the Committee to do, and this also gets to the issue of informing the public and I think following up on Paul's comment of lack of information about whether there really is discrimination, is to consider either holding hearings or town meetings.

I'd actually like to hear a little bit more from Dr. McCabe about how it worked with the Genetic Testing Committee because I see that you held a town hall meeting on the accessibility of genetic testing, and one thing that the Committee might want to think about is serving the role of either convening town halls on particular topics, to get information out to public and to get the public's views about issues, or stepping into areas, such as discrimination in use of genetic testing, to get expert and public testimony to then compile this information and step in and fill this informational void.

The National Committee on Vital and Health Statistics, I know, has sponsored both town halls and hearings on topics, including, for example, the impact of the privacy rule, and we in the Department really use this information to inform us about areas and directions that we need to go in terms of public education. So that might be something to consider in terms of both a method and then identifying issue areas that could be a concrete approach for this Committee.

DR. McCABE: Debra, you want to comment on that?

DR. LEONARD: I'm hearing from a number of sources that more information is needed about whether or not there is discrimination based on genetic testing.

My question is, is that something that this Committee wants to focus on since, in my mind at least, we have the discrimination bill that's moving towards law. I don't know at this point. I mean, the conditions are going to be changing under which there is or isn't discrimination if this bill becomes law, and I don't know if this is a real need and problem area and maybe you can comment or whether this is something that should be more on the back burner and us dealing with issues where we can have a real impact.

DR. McCABE: Yes. We heard from Kim, and I think it's likely, that it will move successfully through the Senate, given the support that it has had and the broad base of support that it has in the Senate. I think we also heard, I know she gave us the caveat that the Senate doesn't necessarily know what's going on in the House, I think from the read that many of us have, not much is happening in the House and the House has not seen this as the same priority that the Senate has.

There's hope that given the majority in the House and given the support of the Administration for this effort, that they might be able to bring it together, but I think many have been skeptical that this is going to make it through this year. I think it's been in process for at least three years. Seven years, seven years. So it's been in process for at least seven years.

So I think that while we're more optimistic than we have been in the past, that's just because it's making some progress this year, but I don't know that it's such a done deal.

MR. MILLER: If I can just jump in, one of the concerns that's been raised around the bill is or one of the issues around the bill is that there really isn't evidence or data that discrimination is currently occurring, and so I think that one of the issues that's going to be thrown into the mix into the House as the House begins to consider whatever bill they begin to consider is, in a sense, is this timely? Is this a problem going on out there in the world?

While there are some cases of great notoriety, including the one brought by my agency, there isn't sort of a landslide of these kinds of cases yet and that, I think, raises two questions. One is sort of to what extent beyond anecdotal stories here and there do you use to build a case or to understand the problem, and secondly, if there are a lot of anecdotal stories but nothing's turning into complaints in any formal kind of way, are there barriers out there that preclude complaints?
Well, either it's because genetic information is so amorphous and it's all over the place, people don't know that that's the reason why they're not getting hired or not getting promoted. People aren't saying we're not hiring you because of your predisposition to cancer. People just don't know why and maybe that's a barrier or maybe there are all sorts of other barriers. People may feel that, gee, this is such private information to come forward and make a complaint, that's going to reveal information about me and my family that I simply don't want to put out there in the public realm. Maybe that's a barrier.

So to get a more analytical understanding of sort of what's going on out there in the world I think would be a useful thing. As Robinsue said, sort of begins to fill the vacuum of sort of hard information that's out there, and I think that's why I'd raise the issue.

DR. McCABE: And I agree that I think that this could be very helpful, if it moves forward and it is successful. Still, frequently legislation isn't perfect and needs to be tweaked and this may help us with that process as well.

DR. LEONARD: So should this go under that first bullet of things that we're going to do to move toward nondiscrimination based on genetic testing? Is this something else under that category of work that this Committee would like to do, that we should try and facilitate this?

DR. McCABE: What I had as a note from the previous discussion, that the Committee staff would confer with the Alliance, EEOC, Labor, NSGC, and others regarding cases of genetic discrimination and any barriers to reporting.

Sarah? So I already had it as a note to myself to talk to staff about this, but we can --

MS. CARR: Well, I wonder, too, about something Paul Miller said about perhaps one of the barriers might be that people don't even realize that the reason they've had an adverse action or something that relates to genetic information, but Paul Zurawski said that you've had 184,000 complaints from people.

DR. ZURAWSKI: On all benefit issues.

MS. CARR: And would that be data to look at?

DR. ZURAWSKI: Yes.

MS. CARR: To scrub through it or work through it to see whether there are some underlying genetic things going on?

DR. ZURAWSKI: Well, we've tried to even break it down, that it would include a genetic classification of complaint. So when our field people who work for the agency receive those kind of complaints, we classify them for our own research and data needs, and so there is some scant evidence of some information that we'd be happy to share with you. It's not as specific as perhaps you would like, but it has to do with more of the timing of where this is going because we couldn't check on rule to see if a plan, a health plan was following the regulation, if that regulation wasn't in force or in final reg. So there was a little bit of that going on, but we do have some real information from 2001.

DR. McCABE: Barbara, would you comment on this topic? Okay.

Just following up with this topic then, Cindy?

MS. BERRY: Paul covered a lot of what I was going to raise, which was, that while in some respects, the train has left the station and so one might wonder, well, why are we going to even tackle this issue of discrimination and analyze it if there's already a bill and they've pretty much taken it as a given that there's a problem that needs to be addressed in some fashion, but on the other hand, if the arguments that are being raised in opposition are really a primary obstacle to House action, then it sounds like there needs to be more than just a letter saying we think this is a good bill, but we could actually help the process along by providing the data.

Now, the old trial lawyer adage of you don't ask a question unless you know the answer could come up as well because if we don't have anything positive to offer, we just have a few anecdotal stories and that's it, in a way, it's reaffirming what the opponents of this legislation already believe. I'm not saying we shouldn't go down that road, but I raise that as something to consider.

My only other question on this topic is what are our capabilities? Is really the best thing that we can do to hold town meetings and gather anecdotal evidence and then talk to the agencies for what they have or is there some -- it almost sounds like there needs to be some in-depth research or a little bit more elaborate study, and I don't know that that's something that we're capable of doing.

DR. McCABE: We can. We can do that. I mean, we can't do in-depth studies. That's not what we have the ability to do, but we can certainly gather data from the agencies here.

Is that fair to say, Sarah?

MS. CARR: Well, sure, and I think we've had the offer of some, but we can't survey. We can't conduct surveys without OMB approval, and so we are limited to a degree in how we can go about -- we can certainly consult with the public and ask for their input on issues and so on, but I'm not sure that if we did that, that would qualify as quantitative data or also be considered anecdotal. So we are somewhat limited, and although I don't know whether the ELSI program has any grants currently that might be looking at this or that might be something to consider, too. I don't know.

DR. GUTTMACHER: If you're asking whether the current grants that are surveying cases of discrimination, none that I know of. I don't believe anybody's --

MS. CARR: Did you ever do that in the past, Alan?

DR. GUTTMACHER: I don't remember ever receiving any applications to do that.

DR. McCABE: Kim, you had a comment on this topic.

MS. ZELLMER: I mean, I would guess from what I'm hearing and sort of my gut feeling is that this is more of a perception problem maybe of people with genetic predispositions than maybe actually a problem, but just the perception, isn't that something that maybe could be addressed by the legislation?

I mean, I don't know. I'm going to guess that most employers are not sophisticated enough to even know not to hire someone because they have some genetic predisposition to some disease. I would guess the reality of it is that probably there aren't that many people that are discriminated against because of a genetic disorder, but I have a feeling that there are probably a lot of people out there who have genetic disorders and they're not disclosing them, certainly to their employers, because they have this perception that their employer may use that as a hiring decision or advancement or something like that.

Just by passing this legislation, perhaps that would give them a little more comfort in getting genetic testing done and getting treatment or whatever that they might need that they're not getting now, and I would guess it's probably more a perception than actual reality where we have a lot of people who are getting fired from their job or not getting a position or not getting advancement because of genetic disorders. I'm going to guess it's probably more a perception problem, but I also think that that's just as real, and if people actually believed that there's a problem, then I think this legislation could certainly help.

DR. McCABE: Someone from the audience, do you have a comment on this? If you could be brief, please?

DR. ROSE: I'm Ann Rose. I'm with Vicro, but what I'm identifying is a previous life. I was at OTA from '81 to '83 and one of the major topics that we worked on -- I don't know how many people know what OTA was, Office of Technology Assessment -- was genetic testing in the workplace, and it seems to me from these discussions that all the issues are still the same regarding discrimination, et cetera, anecdotal data that was there in 1981-83 has not changed. The only thing that's changed is the progress of where we are in genetic testing, and it was the House that commissioned that study to be done. Gore was there at that time, and as a result of not being able to have any more than anecdotal data about discrimination, it went nowhere, and my concern is that will the House again use this as an area to block it versus what you've got going seems to be a train that's running in the right direction and may be helping the House members and encouraging lobbying groups to do that to get the Senate bill through may be a way to consider it.

DR. McCABE: Thank you.

Barbara, and then I want to do some summation and some housekeeping.

MS. WILLIS: You know, as we sit here and wait to talk, everyone brings up what you're going to talk about in the first place. So I guess I'll try to really cut my comments short because so much of it has been touched upon already.

I think my main concern and one of the global topics that I hope that we'll be able to talk about in this Committee basically has to do with access, and again several people have touched on the different aspects under that. Just to enumerate some of them, I think obviously the workforce issues, and I think numbers are important. How many medical geneticists we have are out there, how many genetic counselors we

have out there, and another layer on top of that is the diversity of the people that are providing these services because we know and it has been noted in the literature very clearly that people tend to be more comfortable receiving genetic services from people that look like them or they feel come from the same background and have the same interests, and so I'm not even talking from that strictly from an ethnicity standpoint but also religious standpoints, the levels of ability standpoint, and so I think all of those are issues that we really need to discuss and hopefully come up with some recommendations about.

Another aspect is education of the public and again not just education of certain aspects of the public but all of the public and so putting information on the Internet is not going to do it. There has to be other efforts in addition to that, and I don't know what agency may have already addressed that, and I would find it useful to find any information about efforts that have gone to that.

And then also the physician education again has been talked about very often. I can even tell you in my own practice of prenatal genetics, I still get referrals at 30 weeks for a woman that's 35 and older, that I know has been to that doctor since she was 10 weeks and just for some reason, that referral never got through, and it may be that the referral came through and for some reason, there was a barrier for her to come to me, and I think all of those things we need to try to work on and put some recommendations toward.

And then, I think a new, relatively new or different topic that we haven't talked too much about is minority participation in clinical research, and I think it was even mentioned yesterday that for some reason, minorities aren't interested in clinical research and why is this and trying to identify those barriers.

However, I challenge people that make those statements. At Howard recently, we are still participating in a study about hemochromatosis and iron overload called the HEIRS Study, and each of the clinical sites that were involved in this study were given the charge of recruiting 20,000 participants, and Howard was able to do that along with the other sites with extra efforts that -- I think we learned about a little bit as we got into it and hopefully we'll be able to publish on some of those and get those efforts out there so other people can use them, but I think one of the points that Dr. Reede had made yesterday was to make those efforts on the front end and not just try to catch up later on, and so I think that's just another topic that we can hopefully talk about.

DR. McCABE: Well, thank you.

I want to begin to wrap up the morning and begin to have people prepare for the afternoon. One other area, though, that I want to bring up because Francis encouraged us to think strongly about it but then couldn't be with us today, and Tim has sorted of hinted at it, but Francis was really talking about large population studies and recommendations regarding funding, which is certainly something that we can take up and even though Francis isn't here, I think we need to address that. Is that an area that we want to explore?

We've had a lot of ideas that have been thrown out. We will do the letter to the Secretary. But I want you to be thinking over lunch of what one or two things are we going to pick up as a priority for a product. We're going to have presentations from FDA and the other agencies that we talked about at the next meeting. That's really to bring us up to speed with where things are.

We had talked about possibly another product being a summary by staff of the review with the ex officios and the issues that they brought to bear and certainly summarizing these data in a way that we could all agree on would identify a host of issues and might be helpful to the Secretary and to the other agencies as well in order to understand that. So I would suggest that that might be something we could do and is

really based on work that's already been done up to this point and sort of help us perhaps give us direction for the future.

But I want you to be thinking about what you would like to do over lunch, and then, having said that and given the discussion that we've had, how long do people think that it's going to take? Do you think that we can come to this decision? Do you think it's going to take us till 5:00 or do you think we can come to a decision by a time earlier than that? Any thoughts on these?

DR. LEONARD: Well, when I started my list of priorities, we got through my first two, but my fourth one is the large patient cohort because one of our mandates is research and that is going to be absolutely essential for moving forward the complex disease understanding is having access to that kind of thing. So I know we're not supposed to discuss this now, and I would be all in favor for ending early.

(Laughter.)

DR. McCABE: Okay. I'm seeing body language indicating.

So why don't we aim for wrapping up some time between 3:00 and 4:00, more likely between 3:00 and 3:30, but what this will mean is for people to focus on priorities because we need to leave here with one or at the most two priorities where we can have, in addition to the letter to the Secretary, if I hear no objection, in addition to a summary of how we've gotten to where we are, then really begin to focus on one or two priorities for the future.