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

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

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

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

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

DR. HOOK: Thank you, Ed.

My concern was that all of these issues are important or they wouldn't have made it onto the final list, and I think it's important that we acknowledge that and that subsequent rankings don't diminish the importance of a given issue by our choices. But we also have to recognize on a practical basis that the Tuckson questions are yes/no sorts of issues. Is someone else working on it? Do we have jurisdiction in the first place? I think we should answer those simple questions first just to acknowledge or recognize those areas where we may be able to move on quickly and say it's not that we don't think the issue is important, but for these simple reasons, practically, we probably should move on to something else. So those were the four that I picked out of his list that seemed to be the easiest to answer in a straight yes or no fashion.

DR. McCABE: Thank you.

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

So basically, these are the four points that Chris picked out and mentioned briefly. Does the

government have jurisdictional authority over the issue? Does the issue raise concerns that only the government can address, or would the government involvement be duplicative? We heard today about, for instance, the patent issue may fall into the second. Is there another body addressing the issue or better equipped to address the issue? Again, the same thing with patents. Have the policy solutions to the issue already been worked out?

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

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

DR. McCABE: A Class 2 issue.

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

DR. McCABE: Yes, Joan?

DR. REEDE: Speaking against this issue, the genetic discrimination, I actually think among the 12, when we get through with this, there are some issues that are recurring, and when I think about something like genetic discrimination, it could be addressed in all of the other 11 issues that are mentioned. It should be a part of them. I think to pull out some of them that are overarching issues such that no matter what we did a component would have to address something like genetic discrimination -- for example, if it was determined that a population study was important, clearly genetic discrimination would have to be a part of that. I think what that does, it stresses even more the importance. It's saying it's not something you can pull out, examine once and be done with, but rather it's a recurring theme, that no matter what area we go into, we have to look at what is the impact with regard to genetic discrimination. There may be a few others that fall into that same category of being so overarching that we need to look at them no matter what the topic.

DR. McCABE: Thank you.

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

But with that background, now we will begin an in-depth discussion of the 12 issues with the highest ranked issue, which is access. Can we go back to the other slide? So here we can see that

we have the members list on the left again, the ex officios on the right. We see that access was number 1 on the members' list, number 3 on the ex officios' list. Some of them moved up or moved down. Some of them moved sort of in both directions on the two lists, up on one, down on the other, to bring them a little closer together. I think it will be important to discuss some of these differences as we go through.

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

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

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

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

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

I'll lead off the discussion, and I would say yes, to some extent. Does anybody wish to comment?

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

DR. McCABE: Yes, David?

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

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

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

Joan?

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

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

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

MS. HARRISON: Suzanne?

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

MS. HARRISON: Brad?

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

So my vote is that as a category of one of the things to focus on for the committee, I would say we shouldn't make access really one of them. But when we pick our three or whatever number of things we're going to really focus on, make sure access is clearly dealt with when we talk about them.

MS. HARRISON: Paul?

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

way that I've sort of been prioritizing these things.

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

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

MS. HARRISON: Emily?

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

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

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

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

MS. HARRISON: Emily?

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

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

Debra?

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

MS. HARRISON: So perhaps going back to the suggestion, this can offer as a framework, as a

way to look at other issues. It may be possible.

Cindy?

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

MS. HARRISON: Emily?

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

MS. HARRISON: Hunt?

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

MS. HARRISON: Joan?

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

MS. HARRISON: Kaytura?

DR. FELIX-AARON: Yes, I have two points that I'd like to make. One is on the issue of access. I struggle with the scope of that issue because I find it's so broad. When we try to sort of operationalize it, like Emily and others did, I find that it's even more problematic, because what I hear in the statement that everybody should have it, because equality is a value that everybody should have access, is that it can be interpreted as the fact that access to genetic technology is a human right; however, health care isn't a human right.

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

MS. HARRISON: Robinsue?

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

MS. HARRISON: Ed?

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

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

MS. HARRISON: Cindy?

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

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

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

MS. HARRISON: Martin?

MR. DANNENFELSER: I guess I have a question on access. By access, are we saying really coverage and reimbursement? Is it an economic issue we're talking about, or is it broader than economics? Are we saying that people have access by virtue of having the economic wherewithal to get these tests, and then if so, it seems that that's basically -- maybe that's a

coverage and reimbursement issue. Or is it a broader question as to who will be allowed to have access to this service, and then the question of reimbursement is perhaps an overlapping but not completely all-encompassing issue in that area.

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

MS. HARRISON: Ed?

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

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

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

MS. HARRISON: Joan?

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

MS. HARRISON: Kim?

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

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

DR. McCABE: Well, certainly we can come to the decision. We can have these typed up so that people can see them, but the four categories again would be not high enough priority to pursue in the initial consideration; number 2 is high enough priority but can be dealt with in a brief statement or some other rapid approach; number 3 is that it's really important to every other topic, integral to each of the topics. So in that sense, it gets taken off the table because it will be

included in the discussion of each of the high-priority topics. Then 4 would be high enough priority and meaty enough that it deserves substantive deliberation by the committee.

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

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

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

MS. HARRISON: I agree with you, Cindy. I had the same reaction.

I'm sorry, Sarah.

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

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

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

MS. HARRISON: Is there any opposition?

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

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

(Show of hands.)

DR. McCABE: Anyone disagree?

(No response.)

DR. McCABE: I'm not seeing anyone who is a member who didn't vote, so it appears that that's

unanimous. So then access would move into Category 3, which probably explains why it was number one, because everybody saw it as such a pervasive issue that needed to be considered.

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

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

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

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

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

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

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

Emily?

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

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

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

DR. McCABE: I think it is important to point out that CMS establishes through Medicare reimbursement schedules, what is frequently picked up by the third-party payer. So while I agree

that it's an influence rather than responsibility, but it is an important influence.

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

Oh, I'm sorry. Debra?

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

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

So I think we'll move on to the next question. Does the issue raise concerns that only the government can address, or would efforts be duplicative? My feeling, from what I reviewed, is that our input would be very useful. I don't know if anyone has any other views on that.

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

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

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

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

MS. HARRISON: I apologize.

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

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

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

MS. HARRISON: Emily?

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

things are missing, need to be there for the things that are inadequately described by current codes.

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

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

MS. HARRISON: Ed?

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

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

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

(No response.)

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

(No response.)

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

Any thoughts? Emily?

DR. WINN-DEEN: I think it's been pretty clearly identified that there are certainly some specific gaps with coding for services and testing modes that are just not there, and you're forced to use some sort of make-it-fit generic code which drives reimbursements. So I would say there's at least that level of need for some work, but I'm also very concerned if there's a sort of global

overhaul, as is being discussed, of the whole reimbursement system, that we need to be active participants in that.

MS. HARRISON: Joan?

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

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

(No response.)

MS. HARRISON: No one wants to own up?

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

DR. McCABE: And why was that, Francis?

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

MS. HARRISON: Okay.

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

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

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

MS. HARRISON: Paul?

MR. MILLER: I guess with all due respect, it's for exactly those reasons that I think coverage

and reimbursement should be ranked at the top of the list, because I think the government does have an influence in those issues, not that HHS sort of governs the entire marketplace, but because what HHS does in terms of its government programs is tremendously influential and can be very groundbreaking in setting standards and pathbreaking in terms of what it does. So in terms of policymaking, I think for those reasons this is an important issue. At least it was in terms of our ranking of where there is a gap and where the voice of the HHS Secretary ultimately can be influential.

MS. HARRISON: Linda?

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

MS. HARRISON: Cindy?

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

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

MS. HARRISON: Brad?

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

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

between what gets reimbursed or something that's really helpful, then I'm in favor of it.

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

MS. HARRISON: Ed?

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

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

MS. HARRISON: Agnes?

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

MS. HARRISON: Joan?

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

MS. HARRISON: Debra?

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

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

MS. HARRISON: Robinsue?

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

MS. HARRISON: Emily?

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

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

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

MS. HARRISON: Debra?

DR. LEONARD: Well, in dealing with one private insurance company on reimbursement issues for all of infectious disease molecular-based testing recently, their criterion is is there a published paper out there, or papers, that basically demonstrate clinical utility, and that's their criterion. So walking in with your own information about clinical utility has no impact on insurers to pay for something. It's the published literature currently.

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

MS. HARRISON: Chris, and then Ed.

DR. HOOK: I just want to strongly support Emily's comments. I think that that's a very practical

and important thing, working with investigators and clinicians trying to determine when tests should come out, and that influences when they should be reimbursed for clinical services.

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

MS. HARRISON: Ed?

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

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

Cindy, did you want to comment?

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

(Laughter.)

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

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

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Any abstain?

(No response.)

DR. McCABE: Okay. So that one we'll move to Category 4. Thank you very much, Barbara, for discussing both of those. Number 3, then, is education. Hunt?

DR. WILLARD: This is an issue that was ranked, again, higher by members than the ex officios. I think I can briefly summarize the questions before us in the sense that I think everyone acknowledges there's a gap in genetic knowledge of health professionals. To me, as I read through both the issue brief and thought about it and discussed it with people, there are really two questions to be addressed. One, to what extent is this a federal or an HHS issue, as opposed to that of academic and professional societies and other groups? And the second issue is -- and I always hate to come back to genetic exceptionalism, but this is no exception to that.

(Laughter.)

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

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

Joan?

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

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

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

DR. WILLARD: Other issues? Suzanne?

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

Another factor in this, as you've identified, is that it is our responsibility and part of the focus of HRSA for the diversity of the workforce and having the right health professionals across the country in the underserved areas. So that's another piece of this.

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

DR. WILLARD: Other comments? Debra?

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

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

DR. WILLARD: Chris?

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

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

DR. WILLARD: Francis?

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

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

The organization that hasn't been mentioned yet that I think takes this challenge on as its major

enterprise, if we're talking about other bodies addressing the issue, is the National Coalition for Health Professional Education in Genetics, NCHPEG. NCHPEG has now been around for almost five years. It has, at last count, over 125 professional societies that have joined up to be part of this, representing virtually all of the major specialties and subspecialties of medicine, of nursing, of social work, of dentistry, nurse practitioners, physicians assistants. A long list of those who find themselves in a provider role have become part of this organization.

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

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

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

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

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

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

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

DR. McCABE: So what I'm hearing is that while there are other groups that are involved with this, that there still could be an impact of this advisory committee taking on this topic.

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

DR. WILLARD: Ed?

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

DR. WILLARD: Suzanne?

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

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

Joan?

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

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

Other comments?

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

DR. WILLARD: Yes, correct.

Linda?

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

risk.

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

DR. WILLARD: Thank you.

Other comments on this point? Brad?

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

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

DR. WILLARD: Barbara?

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

DR. WILLARD: Ed?

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

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

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

DR. FEETHAM: Sarah just modified number 2 to say "or through monitoring," and that gives just another perspective on that. It's been identified by Barbara in some of the other discussion.

It's important to keep it on the screen. I think your comment about are we going to learn more through more in-depth study through this group, but to keep it visible, to keep an eye on it, to track it, to keep it on high visibility, again with the role of the federal agencies, plus with this group, with the adaption of 2, if people go with that. I think that's another way of dealing with this.

DR. WILLARD: Yes.

Ed?

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

DR. WILLARD: Emily?

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

DR. WILLARD: Okay, thank you. Sorry. I apologize. I can't see down the line here.

DR. FELIX-AARON: That's okay.

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

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

DR. WILLARD: Thank you for that. Ed, did you have -- Joan first.

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

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

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

DR. WILLARD: Other comments? Ed?

DR. McCABE: Just to clarify, I think the point was that I hadn't heard anyone speaking to putting it into Category 2, but I would agree that I think that makes a lot of sense. Likewise, I think we heard on the discrimination that things may fall into more than one category depending on how we look at a topic. So I think that that seems like an excellent way to approach it, that certainly it could fit into -- what I'm hearing you say, then, is Category 2, to some extent 3, perhaps not as pervasive as some of the others, but wherever there's an opportunity to include it in other discussions, we should, and then 4, that there may be more to do on this topic. Is that correct?

DR. REEDE: Correct.

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

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

(Laughter.)

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

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

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

DR. McCABE: Any further discussion of Category 2?

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

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

Paul?

MR. MILLER: Thank you for inviting me to your sausage factory. (Laughter.)

MR. MILLER: What might be helpful to me, and maybe to the group, I think the people sort of regard education and training as important, but what would be helpful would maybe be to

imagine or maybe spend a moment or two and think out and say, okay, if this was a really high priority, and if we were going to spend the next four months thinking about -- let's say we've dealt with all the other things -- what would this committee do to sort of move the ball forward on it? Would we hold seminars? Would we sort of wag our finger at medical schools? What would we do for this committee to get our hands around education and training?

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

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

Francis?

DR. COLLINS: Well, I think first you'd want to collect the information about what's really specifically already being done. You'd want to have a NCHPEG executive director come and tell you all the programs that they're currently pursuing, what their timetables are, what their success rates have been so far in achieving their goals. You'd want to look at the SACGT's document to see what their recommendations had been, and then you'd try to figure out, as you were asking a few minutes ago, are there gaps identifiable here where this group has the jurisdiction and where something that could be done that's not already under somebody else's purview.

DR. WILLARD: Ed, and then Joan.

DR. McCABE: Joan?

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

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

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

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

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

DR. WILLARD: Agnes?

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

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

DR. WILLARD: Other comments? Martha?

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

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

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

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

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

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

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

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

Suzanne?

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

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

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

Chris?

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

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

Yes, Joan?

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

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

(No response.)

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

aye.

(Chorus of ayes.)

DR. McCABE: Any opposed? (No response.)

DR. McCABE: Any abstain? (No response.)

DR. McCABE: Okay. So Category 2 is where it will go, then. Number four is large population studies. Hunt?

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

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

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

DR. WILLARD: I have no doubt that it's in good hand. (Laughter.)

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

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

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

So the consensus of the group -- and this was a pretty distinguished group, and they came in I think with some skepticism -- was that such a large-scale cohort study would be extremely

valuable. They had no real dissension from that conclusion. They felt also that it would be useful, if possible, to have this across the age range, from childhood to late adulthood, and if you were going to do so, some of the geneticists argued you may as well do this in a family-based approach so that you were covering three or four generations and you had the ability, using the tools that geneticists are familiar with, to be able to test associations and make sure that they're not false positives.

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

So the big issues, of course, that arose out of this were numerous. What exactly would be the study design? What kind of power would you have being able to look at gene/environment interactions for diseases? Of which particular incidence? What could you afford to do? Because the costs would be very substantial and would obviously scale as you went into larger and larger populations. What kind of environmental data could you afford to collect? What kind of genetic data and phenotypic data? How would you deal with collecting clinical information, which in this country tends to be rather fragmentary and non-electronic?

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

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

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

So where this all stands is there have been a number of small follow-up discussions, but the plan really is now to try to formalize that a bit more by assembling a working group of experts to try to flesh out some of the questions that didn't get answered in this rather brief two-day workshop. But this is still very up in the air. It was mentioned this morning, the possibility of asking the Institute of Medicine to get involved in this, and that has not been ruled out. But I think the concerns there were partly cost, which again is a real issue right now, and partly sort of timing

given that IOM studies generally don't happen overnight, and this is a circumstance where if there's going to be some momentum behind this, we probably don't want to have that go on indefinitely or more than it has to.

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

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

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

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

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

DR. WILLARD: Thank you, Francis.

Comments? Debra?

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

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

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

Chris?

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

DR. WILLARD: Emily?

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

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

DR. COLLINS: It's not been fully costed out because so much would depend on exactly what study design is chosen, but it's certainly on that scale in terms of the investment over the course of a 15-year time period. Much of the cost of a longitudinal cohort study, though, hits you up front, because you need to do the accrual phase. You need to get individuals enrolled and collect the clinical information and the biological specimens, and then the monitoring, the follow-up actually tends to be somewhat less expensive on a year-by-year basis.

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

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

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

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

DR. WILLARD: Ellen?

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

The Veterans Health Administration has been actively pursuing a proposal for the type of study that we're discussing here, and that has received conceptual approval from the National Board of our organization, and we're sort of in the final stages of working out some of the specifics in terms of the informed consent issues and the privacy issues. But this would involve collection of environmental data linking to the clinical records system and enrolling on the order of more than a million patients. So that is underway.

DR. WILLARD: Thank you.

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

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

DR. WILLARD: Brad?

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

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

The other issue, just getting back to the whole thing, is that right now it sounds like it's all a big debate about infrastructure and resources, of course, logistics. I'd like to have much more

consensus that the scientific merit is there in having one big group. I mean, today clinical trials are done everyday for drugs, and they don't use one big population. They go pick their populations.

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

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

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

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

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

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

DR. WILLARD: Joan?

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

DR. REEDE: I don't actually think it's the job of our committee to determine the scientific merit or the study design. I think there are much more learned people who will deliberate on that. But I do think that there are things that our committee could look at in terms of equity, in terms of issues such as if we're starting to use convenience data sets, which I would classify things such as

Veterans Administration or some of the other data sets that might be Kaiser or other types of things, what happens to the 43 million Americans who are uninsured and don't fall into those categories who may have very different types of environmental factors or other things going on?

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

DR. WILLARD: Thank you. Emily, and then Ed.

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

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

DR. WILLARD: Ed?

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

DR. WILLARD: Chris, and then Francis.

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

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

Francis?

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

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

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

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

DR. WILLARD: Debra, and then Brad.

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

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

DR. WILLARD: Brad?

MR. MARGUS: So I would be in favor of having this be one of our subjects to cover if the likely result is that we're going to deliberate over whether we're going to make the recommendation that Debra just said. So if we're going to decide whether we're going to endorse doing it or not. That

makes sense. But if it's really for us to go ahead and start discussing all the ELSI issues and access and discrimination, all those issues, it seems a little bit like a waste of time to do it now if we don't even know if this project is going to happen. I mean, are they going to find $5 billion to do it? Maybe by the time you get around to starting it we'll be much more insightful committee members, five years from now or so.

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

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

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

DR. WILLARD: I'm going to call on myself. (Laughter.)

DR. WILLARD: Because I thought I was hearing a reasonable consensus that this was a number 4 issue, and people could then vote among the number 4 issues how highly it would rank, because I would argue that, notwithstanding Francis' discussion of his group and where it's going, there's no guarantee. This isn't a situation that it's either the NIH or it doesn't get done. You could imagine a private consortium getting together to decide to do it. You could imagine one wealthy billionaire deciding that he or she wanted to do this. So I think from our standpoint there are a number of issues that we might address, from the standpoint of genetics health and society, which might add value to Francis' deliberations. It might add value to someone else's thought process on exactly how to go about doing this, whether it's public, private, or somewhere in between.

Chris, and then Cynthia.

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

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

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

MR. MARGUS: That's my vote.

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

MS. BERRY: Just a question to the scientists really of the group. What I'm hearing, and

especially when I listen to Debra, when you speak it almost sounds like this is the equivalent of going to Mars or some sort of massive vision.

DR. WILLARD: It's cheaper.

(Laughter.)

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

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

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

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

Ed, you had a --

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

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

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

DR. McCABE: Debra, you wanted to --

DR. LEONARD: My only concern with making this a 4 is that this committee meets relatively

infrequently, and I would hope that Francis or others for whom this is more of an urgent issue could do the review of is this scientifically warranted, what are the other models that are out there in a more timely fashion. But I may be wrong.

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

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

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

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

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

MR. MARGUS: Second.

DR. McCABE: Okay, Brad has seconded to the motion. Any further discussion? (No response.)

DR. McCABE: Not that we're holding your lunch hostage or anything to that discussion. (Laughter.)

DR. McCABE: Any further discussion? Because we could table this until after the break. (No response.)

DR. McCABE: Okay. Seeing the body language around the table, we'll take it to a vote, then.

All in favor of this as a Category 4, say aye.

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Any abstain?

(No response.)

DR. McCABE: Okay. With that, let's take a 10-minute break to gather the lunches. The lunches for the members and the ex officios are outside the door. They have your names on them. Please pick them up and come back, take a break for 10 minutes, and then we're going to have a working

lunch today.

Thank you.

(Recess)

DR. McCABE: Why don't we go ahead and get started? We still have eight of the 12 to do. We technically have a half hour to do that in, but we want to be sure that everything has adequate discussion. We have an hour tomorrow that we were going to discuss process, but in fact we've been doing that as we go along today, so we can slide over into tomorrow. There was some sentiment that as we got down to the bottom of the list we could spend less time, but that may be because they haven't had adequate discussion here. So I'm not sure that we want to do that.

So we may hold our second straw vote until tomorrow after we've discussed all 12. Does that seem reasonable to everyone? And it's doable, Sarah, with respect to staff needs, to hold the straw vote until tomorrow? Okay.

So, Cindy, this is to discuss public awareness.

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

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

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

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

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

The third question, is there another body addressing the issue, and actually in our issue brief on this topic you can see that there are an awful lot of groups and agencies that are working on

certain aspects of public awareness: the Department of Energy, National Human Genome Research Institute, Health Resources Services Administration and the March of Dimes are teaming up, the American Association for the Advancement of Science, and undoubtedly other groups as well. So the answer to number 3 is yes.

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

The question is for the committee, how much of a priority do we want to place on this? Is this something that we can and should influence? Do we have concrete recommendations that we would like to consider putting together for the Secretary for additional work, work that's in addition to that which is already being done, or is this something that really is in the Category 1 or 2 where it's somewhat important but there are other issues that are more important, or the second category is it's important but we don't need to spend an enormous amount of time working on in-depth analyses and recommendations. I'm not going to put forward an opinion on that one. I just wanted to kick-start the discussion and then open it up for others, and then we can talk about how we can categorize it.

Yes, Agnes?

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

MS. BERRY: Yes?

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

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

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

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

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

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

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

MS. BERRY: Matthew?

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

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

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

MS. BERRY: Linda?

DR. BRADLEY: I think one of the things that concerns us is the lack of balanced information. I mean, the public is getting their information about genomics largely from the media, and that varies from out-and-out hype to maybe overly optimistic, to sometimes dire predictions. What might be more helpful is for them to have a source of balanced information about what makes sense and what doesn't right now, currently, based on what we know, what are the gaps in information. I've heard Francis speak many times about the loss of credibility that we could suffer if we don't start differentiating between what are tests and other genomic applications that

we have some basis for substantiating it should be in practice and others that are being offered -- and we've all seen some of the websites -- where there really aren't data available.

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

MS. BERRY: Emily?

DR. WINN-DEEN: I thought SACGT was working on some kind of a patient or something brochure, and I think it might be helpful if we could also, as you mentioned before, get a copy of that to see what is happening. I guess I interpret patient awareness as much broader, just really making sure that all the education is there through the school years, but I'm particularly concerned about people who have completed school, and there's a lot of them out there, and how are we going to make sure that they're getting correct information and not just reading about it in whatever news source. What's on the Discovery Channel or PBS are probably good programs. There's probably just as many bad sources of information out there, and how do we help people know where to go, what to do.

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

MS. BERRY: Ed first, then Martin.

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

MS. BERRY: Martin?

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

MS. BERRY: Hunt?

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

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

websites than they will to other websites.

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

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

MS. BERRY: David, then Ed.

DR. FEIGAL: There are two layers to think about, about public access to your opinions. One is as experts on genetics, and that is very broad. But the other is the work of the committee itself. This is a public committee, and although there is a small audience in attendance at many public committee meetings, the ability to actually have an open and transparent discussion of controversial issues and get them in front of the public is something that's very unique in the United States. We're very aware of that at FDA because our advisory committees are more heavily covered sometimes than our own decisions on the same products.

But it brings the debate to the public in a way that doesn't happen in Europe, for example. In Europe, the decisions are just made, and even the basis for the decision can't be reached by anything like FOI.

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

MS. BERRY: Ed, and then Emily.

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

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

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

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

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

Now, maybe there is a genetic component, but until that's proven and well documented, we don't want people getting either the wrong health care or not getting the right health care because they don't really understand what's going on and what's true and what's not true. Now, a large part of that should be happening with a health care provider. But as you mentioned, your email box is filled with misinformation, particularly around weight loss and that area. I think if we're not careful about educating the public so they really at least know where to go -- I mean, a public awareness campaign can be NIH has this website. It doesn't have to mean that you do all the in-depth education in your primary format.

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

MS. BERRY: Francis, and then Joan.

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

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

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

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

likely then to ask their health care professional.

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

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

Genetic testing could help you? Well, that doesn't sound so good. Know your family history. That would be a good thing for people to know about. Maybe that would be a decent message, but it would be a little hard to get a lot of people to jump up off their couch about that one. What is it that we want people to be aware of, and what actions do we want them to take? I don't think we're even quite in consensus about the answer to that question, which makes it hard to design a public awareness campaign.

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

MS. BERRY: Joan?

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

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

MS. BERRY: Yes, Kimberly?

MS. ZELLMER: I just wanted to reiterate what Francis was saying. I think that when we're talking about public awareness, I think there's a lot of information out there, and I think that part of it is what the public is interested in. I think that personally, I'm an educated person but I knew very little about genetics until my daughter was diagnosed, and that obviously generated an

interest, and I found plenty of information once I wanted the information. I think it depends on what specifically you're trying to raise awareness with the public, because I think if you're just talking sort of general genetic principles, those that are interested have plenty of resources to go to and look for.

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

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

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

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

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

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

MS. BERRY: Martha?

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

MS. BERRY: Brad, did you have something?

MR. MARGUS: I'm not really sure how we can improve awareness. I like the idea of using our

pulpit as a committee, anything we decide to support like a large population study, to use our opportunity to shout it from the mountaintops.

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

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

DR. McCABE: We're going to need to wrap this up very quickly so that we can move on to the public comment.

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

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

Kimberly?

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

MS. BERRY: Yes, Barbara?

MS. HARRISON: I think I'd just add a thought that's been going through my mind, which is really how pervasive and how important public awareness is. I mean, the reason why we don't

have underrepresented populations participating in studies is because they're not aware of genetics. They don't know how important it is for them to be involved in that clinical trial or for them to be involved in this research protocol, because it significantly contributes to the development of drugs and that kind of thing.

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

MS. BERRY: Yes, Joan?

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

DR. McCABE: Okay. Well, having heard the discussion -- thank you very much, Cindy. Having heard the discussion, do I have a formal motion? It sounds to me like this is moving toward a Category 3, transcends all issues, needs to be included in any discussion of any issue about how to make the public aware of that. Can I have a formal motion to that effect?

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

Any further discussion of this?

(No response.)

DR. McCABE: All in favor, say aye.

(Chorus of ayes.)

DR. McCABE: Opposed, nay?

(No response.)

DR. McCABE: Abstain?

(No response.)

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