Discussion of Draft Vision Report
Facilitator: Emily Winn-Deen, Ph.D.

It leads nicely to the next issue on our agenda. In March, we decided to prepare a summary of our systematic prioritization process in lieu of a vision report. Though the charter describes a vision and role for this committee, we thought that a summary of our process and the issue briefs themselves would be of use to the Secretary.

I'd like to again thank Emily Winn-Deen for her leadership throughout the prioritization process and her assistance in developing the draft vision report, and I'll now turn to Emily to facilitate the discussion of the report.

DR. WINN-DEEN: Thanks, Ed.

We have to be clear in acknowledging who really did the work on this report, which was the staff for this committee, specifically Fay Shamanski led that effort, and I want to acknowledge her efforts and the fact that she was a very apt ghost writer for the committee.

What I'd like to do this morning is to just sort of go through what I believe is pretty much just a recap of our discussion at the March meeting, make sure that everybody has had a chance to look at this summary, agrees with it. If we have any discussion items, we should put them on the table now because I know that one of our goals is to put this whole set of issue briefs, as well as the summary, up as a public document and one of our work products.

So I guess the first thing I'd like to ask if just if everybody has had a chance to read it and agrees that the written summary is a correct representation of our thought process and our end conclusions from the discussion. I know I had a chance to do a little bit of proofreading, but I don't know if there are any other comments that people would like to make at this time, so I want to do that first.

Hunt?

DR. WILLARD: A point I've made before, as well. I think it's important, especially for written documents that come from this committee, that even though our name is the Advisory Committee for Genetics, Health and Society, that we get the word "genomics" in there as well, at least periodically in executive summary sections, because there will be some of the audience who will think that what we're saying is relevant to genetics, i.e. the last 20 years, but not necessarily genomics in the next 20 years. I know what we all mean, but I think we should be careful in choosing language that conveys that to our audiences.

DR. WINN-DEEN: So would you like to see everywhere that it says "genetics" changed to "genetics and genomics"?

DR. WILLARD: I wouldn't do it everywhere because that will get tedious to the extreme. But I think even in that very first sentence, for example, it would broaden the sense of that sentence and not hurt it a bit to say "advances in genetics and genomics promise to improve human health," et cetera. Just picking a few spots throughout, especially in summary sections, I think could have effect and be more inclusive.

DR. McCABE: We can even change it in the title to genetics and genomics so it's right up front, "Toward a Vision of the Integration of Genetics and Genomics in Health and Society," if that's acceptable to the committee.

DR. FELIX-AARON: I have a question.

DR. WINN-DEEN: Yes.

DR. FELIX-AARON: Why would we keep both? Why not just genomics? What do we lose by dropping genetics?

DR. WINN-DEEN: I'll give you my answer to the difference between genetics and genomics.

DR. FELIX-AARON: Sure.

DR. WINN-DEEN: Genetics looks just at germline DNA, whereas genomics

can also encompass expression analysis, and they're quite different things. So one is what's the basic program, and the second is how is that program expressed at different points in different disease states.

DR. FELIX-AARON: Right. But for the purposes of this group -- I know those are the technical differences, but for the purposes of this group and the work that you do, I mean, I'm asking the question to the group, what would we lose by instead of having genetics/genomics, focusing on just genomics?

DR. McCABE: Yes, I also think that the practice, the clinical practice of this discipline is genetics, as opposed to genomics, which I think of more in the analytical side. But the medical practice is the practice of genetics. So taking it a little bit further, I think it's a subtle difference, but I think that if we want to have credibility within the genetics/genomics communities, we need to try and use the two terms. If we went just with genomics, I think it would be leaving behind the medical practice and some of the issues about germline inheritance.

DR. WINN-DEEN: Okay. So then I think the next thing that we wanted to do was to go through whether the report actually accurately represented our whole process for going through a large set of possible topics and the voting process and triaging and prioritization process that we went through with the main committee, with the ex officios, and in the course of our discussion at the March meeting. So I just want to ask if there's any discussion or if anyone feels that we've failed to capture that in an accurate representation.

(No response.)

DR. WINN-DEEN: Wow. I'm really excited, because after we had all that discussion on whereases yesterday, I was afraid that this might take a really long time.

So I'll take the silence as everybody's ascension that this is an accurate representation of the process and the conclusions of that process.

I guess the third thing I wanted to do was to, if we feel that the overall summary report, "Toward a Vision of the Integration of Genetics and Genomics in Health and Society," is a good representation of our thought process, I just want to give people a chance if there's any comments on any of the issue briefs, which I think we had a few comments on at the last meeting, and I think most of those comments have been incorporated. But if anyone has any further thoughts or corrections, comments, whatever, on any of the issue briefs, I'd like to open those up as well for any recommendations.

Ed?

DR. McCABE: Before moving on to that, we were just having a little bit of a sidebar here, and one of the things, because of this discussion about genetics versus genomics, that we might try and do is a fairly brief glossary of some of the key terms that we could work on, if that's acceptable to everyone.

DR. WINN-DEEN: Yes. Clearly, I think that would be a useful -- I don't know if it needs to be in the body of the Towards the Vision statement, but just as part of the appendix would be a glossary of terms.

DR. McCABE: Right, and it's not going to be an extensive 30-page glossary, but picking up the key terms like we just discussed I think would be important for people to understand what the real issues are here.

DR. WINN-DEEN: Yes. I actually think that there's a lot of those things that the previous committee already worked on, good definitions, and so we can just pull those together.

DR. McCABE: Yes, that's where I was really thinking we had those glossaries probably largely in hand and can extract from them.

DR. WINN-DEEN: Any dissenting thoughts on that?

(No response.)

DR. WINN-DEEN: Okay. So let's ask staff if they would add that to the appendix as well.

Can we turn to the issue briefs?

DR. FEETHAM: Not to do revisionist history, but in the bullet on the first page of the summary, my recall from the discussions on the third bullet that we talked about, again to be consistent with the name of this committee, it's the ethical/legal/health, which to me is broader than medical. Also there was, as I recall, part of the discussion on the large population studies was the economic. I just bring that up because I think that's what I recall from our discussions, which is broader than what that third bullet is.

DR. WINN-DEEN: Right. So economic, you're talking about sort of who would fund the large population studies?

DR. FEETHAM: Well, I thought that was part of the implications. It's effect on health, but also there's the cost benefit, which was part of the discussion as I recall it, and that was my thinking on it.

DR. McCABE: Just so we have it for the record, investigations of the ethical, legal, health, economic and social implications, is that the way it would read? Or maybe we could put health first, Sarah is suggesting, which I think is a good idea.

DR. WINN-DEEN: Thank you. It's good to have a few comments. I'm underwhelmed by the response right here.

I think that these issue briefs are also quite important for us to have officially blessed as part of this committee, because they are going to in some ways frame the issues, but also direct the way that we think about some of the ones that we have put on a prioritization path. So I'd like to take comments on that.

Deb, if you have some?

DR. LEONARD: In just looking at the bullet points, also "enhancement of oversight of genetic technologies and services." This makes it sound as if there's a need for enhancement, as opposed to assessing the oversight. I mean, it's put in almost a negative light, and we did hear from CLIA and FDA and CAP. There is a lot of oversight of genetic testing services now. So could we change enhancement to assessing the oversight? Because that's really the process that we did. We looked at what was being done and not necessarily enhancing it but evaluating.

DR. WINN-DEEN: So would you say assessment of the need for enhancement, or should we just say assessment?

DR. LEONARD: I think what we did was to assess the oversight. It's just that enhancement makes it sound like there's a deficiency in the oversight of genetic tests, which I don't think currently exists for most areas.

DR. TURNER: Except that isn't this where we got into the testing that you can order online by sending in mouth swabs, and we had that discussion about all the different ways that maybe we wouldn't bless as official testing but that the community at large sees as an opportunity for testing?

DR. LEONARD: But that's the final bullet, assessing the pros and cons of direct-to-consumer marketing of genetic tests. I think that's been separated out as a separate bullet. It may be a fine point, but just looking at how all these others are stated, this is stated in such a way that it sounds as if there needs to be more oversight, rather than we are assessing whether the current oversight is adequate or not.

DR. WINN-DEEN: Right. I think that's fine.

Fay, can you make that change? Okay.

Going once, going twice -- we've got another red light on over there.

DR. SEMERJIAN: I have not been involved in these discussions. This is my first time here. I noticed that in Dr. Murray's presentation, he added an item into his presentation that was not in the printed version about the quality assurance of genetic testing, et cetera, which sort of relates to the same issue.

Perhaps the sensitivity here is with regard to the word "oversight," no?

Because I think that there is room for improvement in terms of the quality of measurements, the traceability of measurements to national standards, et cetera, because I think this is a very different testing issue with regard to genetics versus the run of the mill clinical measurements where we have our cholesterol tested many, many times, whereas genetic testing perhaps will be done once in some cases, and you rely on that information for many decisions, that you need a different level of quality assurance, reliability of those measurements.

I'm not sure that we are at that point. I thought this was perhaps part of it, but I thought maybe the issue was do we really want to say oversight, or do you want to say enhancement of quality assurance measurements or quality assurance efforts or something like that? But I think there is room for improvement in that regard in terms of quality assurance.

DR. WINN-DEEN: Ed?

DR. McCABE: What if we state it as "assessment of quality assurance and oversight"? Would that be a way of getting that point in there? Because I think that was part of the discussion in point of fact.

Sarah is commenting that does this get to clinical validity, or is quality assurance more analytical validity?

DR. WINN-DEEN: Well, I think quality assurance is more getting at whether the answer you gave, given the analyte you tested, was the correct answer, which is different than whether that answer has any medical utility. I think they're two separate things, and I think the point that was just made by NIST is that because things are potentially once in a lifetime tests where you don't have a chance through repeated testing to catch an error, do we have some higher obligation to provide QA/QC kind of mechanisms to assure that the test result is actually a correct result.

DR. McCABE: I'm just trying to figure out how to word it to assist staff. What if we don't make quality assurance, if we leave it at quality, assessment of the quality and oversight of --

DR. WINN-DEEN: I think it might be useful to actually look at the issue brief that goes with that bullet point, which is in the appendix "Oversight of Genetic Technologies Issue Brief." I think there's a lot of discussion about who has oversight responsibilities, what the current status is, but there's not really a separate discussion in that brief on QA and standardization of methods as much as just in the current medical system what groups are responsible for trying to provide the oversight that would be relevant for genetic tests.

Cindy?

MS. BERRY: What if we just said assessment of oversight, blah blah blah, and refinement where appropriate, so we aren't really making a judgment about whether we're definitely going to improve something or that there's something in need of improvement, but we're recognizing the fact that we're always going to have to refine things given changing circumstances. I don't know if that does the trick.

DR. WINN-DEEN: I'll turn to the people who brought up the question. Ed, if you want to comment.

DR. McCABE: Well, I was just looking, and if one reads the issue brief, the issue brief pretty clearly says need for enhanced oversight of genetics tests and leads logically to that point also, issues about protecting the public and access to new and cutting-edge technologies. But I think the way it was stated, perhaps the way you originally said it, assessment of the need for enhancement of oversight, since that clearly is in the issue brief, and by stating it that way it doesn't presume a conclusion.

DR. WINN-DEEN: Okay.

Hunt, you had some comments?

DR. WILLARD: I was just going to point out that in an executive summary like this, all of the other terms are very neutral. They don't tip our hand one way or another, we just list opportunities. So in that sense, Debra's point is absolutely right on target. I don't think

we have to sort of lengthen the bullet point to cover all contingencies. We're simply saying we were evaluating it and read further if you want to know what we decided. In that sense, I'd tip more towards neutral terms as much as we can.

DR. WINN-DEEN: So assessment of the need for oversight, or need for enhancement?

DR. WILLARD: I don't think we want to say assessing the need for oversight. That would suggest we actually consider the possibility no one needs to have oversight.

DR. WINN-DEEN: So assessment of the need for enhancement of oversight of genetic technologies and services?

DR. WILLARD: I'm not a big fan of the word "enhancement." I think I'm with Debra on that one.

DR. WINN-DEEN: Okay.

DR. LEONARD: Actually, if you look at the brief, while the next to the last paragraph says "While there seems to be a consensus about the need for enhanced oversight of genetic tests," the beginning of the next paragraph states the question "has a balance between protecting the public and access to new and cutting-edge technologies already been achieved? Do current regulatory mechanisms strike an adequate balance between access, safety, competition and independence of medical practice?" So those are the questions that we're looking at. I mean, that statement at the first sentence of the previous paragraph is pretty strong, actually, and I never really caught it before this point, because that's saying that there is a need for enhanced oversight, whereas the questions that are being asked at the end are really more balanced.

So I would just be happy with changing it to "assessment of oversight of genetic technologies and services," or "assessment of quality and oversight of genetic technologies and services," because it is, as Hunt points out, more neutral and consistent with the other bullet points as something we're going to look at, without a pre-conclusion about what needs to be done.

DR. WINN-DEEN: Okay. So I'm going to ask -- oops. We've got Judy Yost.

MS. YOST: Yes, I just have a comment, and maybe it's a very subtle point, because I think we had this discussion once before about the terminology when referring to this. My concern again, and maybe it's not legitimate but I think I need to bring it forward, is that that assessment, just leaving it as is, I realize your need for neutrality in an executive summary. However, when you're saying that, it sounds like an active ongoing monitoring of what the oversight is, like you're actually doing -- you know what I mean? -- taking an active part in that oversight, and I don't think that's really what's intended as the role of this committee, frankly.

DR. WINN-DEEN: I think what we decided as a committee was that we would try and just keep this on the radar screen so that we would have periodic reports from the different bodies like CLIA about where things are.

MS. YOST: And that's fine, absolutely. But at that level.

DR. WINN-DEEN: Barbara?

MS. HARRISON: I was thinking maybe a compromise would be to say "assessing the current state of oversight" or "assessing the state" or "the status," so you imply that it's just a one-time look at the topic.

DR. McCABE: And since we were planning to monitor occasionally, as we concluded, I think assessing the status rather than current status would be the appropriate thing.

DR. WINN-DEEN: I'm going to ask Fay to read back what she thinks that bullet point says now so that we can see if we have consensus.

DR. SHAMANSKI: "Assessment of the status of quality and oversight of genetic technologies and services."

DR. WINN-DEEN: Okay. Is everybody happy with that as the bullet?

(No response.)

DR. WINN-DEEN: I'll take silence as a yes. Quick, turn off all the mikes. Joan?

DR. REEDE: In another area, looking at the issue brief on genetics education and training, and given our long conversations yesterday, it really doesn't address issues of diversity in the workforce or cultural competency, and I think those words need to be incorporated within the issue brief.

DR. SHAMANSKI: Do you mean within the issue brief or within the report itself?

DR. REEDE: I think at least within the issue brief, so that when it asks about adequacy of the genetics workforce, there's no mention of diversity there, there's no mention of cultural competency.

DR. WINN-DEEN: So, Joan, do you want to suggest a place where that might go? Should it go somewhere in the first paragraph?

DR. REEDE: It could, it you look at the next to last paragraph, it says there are questions about the adequacy of the genetics workforce, and then it speaks about specialists and generalists, and I think there to talk about adequacy and talk about adequacy in terms of representation and diversity of groups would be a logical place to put that.

DR. WINN-DEEN: Any dissenting votes on adding those two points to the workforce issue brief?

(No response.)

DR. WINN-DEEN: I think those are really good points and they clearly were brought up as important parts of the training and education of our workforce that's going to be dealing with genetic issues in the future.

Are there other comments on issue briefs that we should take up? Fay?

DR. SHAMANSKI: I just wanted to point out that we did get some public comments on the issue briefs, and I think the committee needs to talk about whether we're going to send those out for public comments. I just want to remind you that when we wrote them, it was just to present a balanced view of the issues, not to present the committee's position on the issues but rather just to give all the background information on which to base your decisions. So the question is whether you want to send those out for public comments and whether we want to change them further, or do we want to keep them in the current state that we had determined previously?

DR. McCABE: In the table folder is the response from AHIP. So America's Health Insurance Plans basically says "We believe concerns" -- I mean, there are a number of things here, but I'll summarize. "We believe concerns about possible genetic discrimination by health insurance plans are largely unfounded." It continues on. Sarah is stating that they point out factual disagreement, and probably we should restate how we spoke of ERISA. It says, "We would also note that the discussion on page 3 of the genetic exceptionalism issue brief incorrectly states the impact of ERISA on state laws dealing with genetic privacy and genetic discrimination and health insurance and employment. ERISA does not apply to insurers or to health information privacy employment. Rather, ERISA is a federal statute that governs pension and welfare benefit plans, including health and disability, income benefit plans." So we can make that factual change.

DR. WINN-DEEN: Right. I think what we should do with the public comments -- this is my personal opinion, but we can get a committee consensus -- is to take the places where we've gotten comments like this that are related to factual things and just do a fact check, because obviously you drew your information from some source and they're drawing from some source, so let's try to fact check it and not just blanketly take the public comment as the correct information but do our due diligence on that, and then I think that would be highly appropriate to make any factual corrections.

I guess from the point of view of the issue briefs, I agree that the point of

them was to try and just give a balanced view of what the issue is and what sort of the things are under discussion or that might require further discussion and to try to make sure we've captured those things as well, without drawing a committee conclusion in any way. I think these were intended to frame the issues so that we could then go through each issue and, as we did yesterday with the education, now we're going to make a specific recommendation in our resolution about what we believe should be done, and I think that's completely different than just to frame the issue brief, which is what these were intended to be.

DR. McCABE: I think clearly from discussions that have been held at every one of our meetings, "the concerns about possible genetic discrimination by health insurance plans are largely unfounded," I think this and the previous committee has disagreed with this, the public disagrees, and we in fact will try to bring the public to the next meeting to discuss that where there has been discrimination.

DR. WINN-DEEN: Well, I think it's completely legitimate to say that there is a debate on whether or not it is or isn't that certain stakeholders feel there's no need for legislation because there really haven't been any major abuses, and then there's other lines of evidence that say despite the fact that maybe there haven't been very many highly publicized cases of abuse, there still is when you do public opinion surveys a feeling among the public that there's a fear. So how do you resolve that? That in itself is a dilemma. How do you resolve the issue of overcoming public fear when there's not actually too much documentation that that fear has a rational basis? But we still have to deal with it. It's still a barrier to the implementation of this and to the practice of medicine.

DR. WINN-DEEN: Other comments on any of the issue briefs? Hunt?

DR. WILLARD: It might be, since this is intended to become a public document, that at the beginning of the appendix where it simply says "Issue Briefs on 12 Priority Issues," that we add a brief paragraph explaining what we mean by issue briefs to essentially argue what you just said, that they're not designed to give an answer, they're designed to lay out the issues for the committee, and by extension for the public, so that people don't read these and believe that we're somehow either trying to make a recommendation or refusing to try to make a recommendation.

DR. WINN-DEEN: Right. That's something staff could do, just write a little summary paragraph with that.

DR. LEONARD: That is kind of explained at the top of the briefs, though, in that statement that's on every page. So something similar to that put at the beginning in bigger letters so you notice it more.

DR. WINN-DEEN: Yes. We just want to make sure that we capture that these are intended to frame the issues rather than reflect any statement of what this committee has arrived at as a conclusion on that particular issue.

DR. McCABE: I would point out, though, that if we finalize it here, then it does become the official view of the Secretary's Advisory Committee on Genetics.

DR. WINN-DEEN: Right, but it's a view basically that this is an issue and here are the questions that remain to be answered, rather than here is our recommendation for what to do about it, which I think are quite different things, and I don't think, for most of these issues, we're quite ready to put our stake in the ground and say this is it, we know exactly what we want to do, go for it. We're framing what we want to do for the next couple of years of this committee's life.

DR. WILLARD: It's particularly acute when you go to the vision statement issue brief. I mean, most people would go there thinking, ah, this is where I'm going to see the vision. In fact, what you get is a lot of questions that say should we have a vision, should we ask someone else to have a vision, and it actually doesn't declare the vision. So I think it's important that people know what these are, and in particular what they're not.

DR. WINN-DEEN: Other discussion from any of the ex officios?

Muin is reaching.

DR. KHOURY: I'm not sure if what I'm going to say might sound a bit too harsh. I missed the last meeting, so I'm not sure how you got to where you are right now. But I think this committee should be more bold, or bolder, and I agree with Hunt here. I was reading the document here, and it meanders along. I think you need to be bold and establish a vision for how this stuff is going to happen. I mean, people know about the issues related to genetics. I was hoping my comments this morning might elicit some reaction, but I guess we all ate too much protein last night.

So just to encourage you to be bold. I think the country needs help in using genetics to improve health and help society. If this is the place to come, just be bold.

DR. McCABE: I would just point out that, as Hunt stated, these really are briefs that we then prioritized in terms of how we would move forward. So they were not the answer; they were the beginning. I would argue that it's not that we don't have a vision, that we voted not to have a vision, but we voted that perhaps that's not where we should spend our time in crafting a vision, but rather to get down to some specific approaches.

DR. WINN-DEEN: My recollection on the ranking of these various things was -- since I put the vision statement on the table, I sort of paid attention to how it fell out in the ranking, and it didn't get ranked very highly by most of the people who voted on the issue. I think people were more concerned with us spending our time on the actual issues than spending however many hours writing the vision statement, which we could have potentially gone to the "Dilbert" vision statement builder website and picked out the right keywords and had one created for us.

So I think our vision is going to come from the issues that we've chosen to prioritize and trying to take specific action on those issues. So being bold in a specific way rather than in a more general way.

DR. LEONARD: Is "vision" the right word? I just wonder if "vision" -- I mean, maybe this is more our roadmap rather than our vision, because we're going to be creating the vision of what we want to do and actions to take and things as we work through these different issues. This is not really -- you're right, one of the issues was to create a vision statement. What we ended up doing was creating a roadmap.

DR. WINN-DEEN: Yes. So I would be okay with a shift in the summary brief to roadmap instead of vision.

Hunt, let's hear from you.

DR. WILLARD: Two points. One is the unfortunate confluence of terms. So we end up with an issue brief on a vision statement, conclude that we aren't going to spend our time on that, and then write a document that's entitled "Towards a Vision," suggesting that despite our vote we're going ahead and doing that. So some post hoc changing of terms may be of some value.

The other specific suggestion I would have, just again to remind readers and to avoid potential misinterpretation of what these issue briefs are, is perhaps put as the last entry for each of the issue briefs committee outcome or something like that, that reminds the reader how this was then prioritized. This issue became one of the top priorities for 2004, or this issue was determined to be an overarching issue. Then under the vision one it could say what we just all said, that rather than having the committee deliberate on the need for a guiding vision statement, the committee instead decided to do something else. It just would clarify and allow readers six months later to actually understand what thought process we went through.

DR. WINN-DEEN: It might actually be worthwhile, Fay, even ordering the issue briefs instead of alphabetically at this point. I mean alphabetically was completely appropriate when we hadn't ranked them, but maybe at this point it would help also even just in the Table of Contents and in the order that they are presented to the reader on the website or whatever mechanism to put them in the priority order that we have established now so it's very

clear to someone scanning it what the priority is. Then our minutes, as we go through our subsequent task force meetings, will have I think a real clear way of capturing, okay, we said these were the top priorities, they were presented at this meeting, the outcome was a specific recommendation. Coverage and reimbursement we obviously have decided is going to take several meetings before we're ready to put out a committee statement.

But others, we hopefully at the end of this meeting will have two committee resolutions. I think that really will also help clarify to people who are following our progress that the priority issues are being dealt with and that there are outcomes.

Hunt?

DR. WILLARD: In terms of the summary statement, there actually are five categories -- we took these 12 potential priorities and put them into five different categories. So I have two suggestions. In the summary statement, I'd actually divide them up, be a little more telegraphic and divide them into the five sections, use bullet points, help the reader understand. That's what an executive summary is supposed to be for. Then I would agree with Emily to actually then organize the Table of Contents into those five categories. Within each one you can alphabetize. But then it's very clear where one is in terms of those five.

DR. McCABE: So just to help staff, we're on the outcomes section, and I'm looking for it, under Tab 2.

MS. CARR: I was just raising a question about the five categories. We started with four, I think, and then --

DR. WILLARD: Then we have to rewrite the executive summary because that paragraph divides into five. So you've got two issues that are the highest priority, then you have two other issues that are -- sorry, three other issues that are "undertaken" for exploration. Then we've got two others that are short-term action, two others that are monitoring, and three others that are overarching.

DR. McCABE: So rather than the way we had set up the prioritization initially, we could take that penultimate paragraph of the summary and just make it a little easier to discern what the real outcome of that discussion was.

DR. WINN-DEEN: I think just some bullet points and pulling things together so that it's all internally consistent. So the way we categorize things, they're listed that way in the Table of Contents, they're listed that way in the body of the summary, and then they're subgrouped that way in the appendix as well.

Ed?

DR. McCABE: Just to go back to the point about vision being one of our lower priorities, and yet the title of the document, what if we made it rather than "Toward a Vision," which is how we got there without being a vision statement, and I think that was the subtlety, but if we made it "Mapping the Integration of Genetics in Health and Society" or "A Roadmap for the Integration," I was thinking with the mapping being a little bit of the genomic allusion there.

DR. WINN-DEEN: So the question is can we steal Muin's CDC little road logo in helix form.

DR. KHOURY: Public domain. Go ahead.

DR. McCABE: So "Mapping the Integration of Genetics" --

DR. WINN-DEEN: And genomics.

DR. McCABE: And genomics.

DR. WINN-DEEN: I actually like the integration. I think "A Roadmap for the Integration of Genetics and Genomics in Health and Society" or something along those lines would be --

DR. McCABE: Okay. We just want to get it specifically.

DR. WINN-DEEN: Yes. I'd just like to keep the word "integration" there because I think that's really the key thing. There are a lot of activities, and they need to all be

somehow coming together in confluence so that this can actually happen.

DR. SHAMANSKI: Could you just review for me what you decided on for the title? Sorry.

DR. McCABE: I think it is "A Roadmap for the Integration of Genetics and Genomics in Health and Society." Is that correct?

DR. WINN-DEEN: That was my recommendation. Is everybody okay with that?

MR. MARGUS: You're going to call it roadmap? I mean, it's not the roadmap. It's developing a roadmap where the prioritization of the issues now have to be reviewed in order to come up with a roadmap. But it's not actually the roadmap, is it?

DR. WINN-DEEN: Comments?

MR. DANNENFELSER: I recommend going back to mapping. I think it covers that concern.

DR. LEONARD: Brad, why has a roadmap not okay? Because we've set out where we're going to go, and then we're going to go down the road to do it. So in that sense, this is our roadmap of what we're going to be working on.

MR. MARGUS: I see, a roadmap for us. I get it. I thought you meant a roadmap as in the way --

DR. LEONARD: Well, I see it as a roadmap for us, for the committee.

MR. MARGUS: Okay. Yes, I cave completely. Use it.

(Laughter.)

DR. WINN-DEEN: So do we need to say "SACGHS Roadmap"?

DR. McCABE: The subtitle is "A Report of the SACGHS," so I think that's clear.

DR. WINN-DEEN: All right. Now, don't feel compelled to fill the time just because there's time.

DR. McCABE: We have plenty of things to do today.

DR. WINN-DEEN: Yes. I want to make sure we've captured everybody's comments, but we don't have to sit here and wordsmith minutiae just to fill the time until 1 o'clock.

Deb?

DR. LEONARD: Just one quick point. "Health care" is sometimes hyphenated, sometimes split. Could that be consistent throughout?

DR. WINN-DEEN: Agnes?

MS. MASNY: Just a question. Did we finalize the question we had earlier about having this go to the public for comment?

MR. MARGUS: I'll echo what I think we already heard, and that is we spent the last year only trying to go through the issues to figure out which issues we now want to focus on. If someone wanted to make public comment, it wouldn't be about any stance to take on those issues but about whether those issues are important for us to then pursue. While I guess they could still make a comment on this not on particular issues but about whether those issues are important or not, they have had the last year to do that, and we've heard from a lot of public comment. So it doesn't appear to me that we need to do that now.

DR. McCABE: And I was going to ask that at the conclusion of this session we then make a decision -- I was going to propose that we make a decision to finalize the document with the changes recommended and that be the finalized document. It will then go public. Obviously, the public can make comment then. But it's more, I think, as Brad has suggested, it will be comment to help guide us in our future deliberations on these issues. But in terms of what this document sets forth to say, we've had it out there, we have voted upon it. People may disagree with our prioritization, but I think it would be important to move this forward to the Secretary.

DR. WINN-DEEN: Right. So I would agree with that. I think that we certainly are interested, always interested in comments from the public on any of these issues. As I mentioned before, I think the important public comments to incorporate here are the factual comments so that we're not misstating facts. But at this point, unless there's some other aspect to an issue that we haven't considered, I think that would be a legitimate thing to add to an issue brief, if there's one more question that should be on that list of questions to consider. But beyond that, I think we should try to finalize these today and then seek the public comment as we get to each issue and really want to delve into it and get all the public input on that issue prior to making a specific recommendation by the committee.

Ed?

DR. McCABE: Part of my concern is if we consider the calendar, it's important to move this forward with some dispatch.

MR. DANNENFELSER: At the risk of wordsmithing, just a small point on page 5 of the roadmap. The top paragraph is a reference that says "CLIA-certified laboratories." I think that's the first reference to CLIA that I think we could use the full name there.

DR. WINN-DEEN: Right. I think part of the appendix of definitions might be also just a listing of all the key acronyms and what they actually mean.

DR. McCABE: That could now make it 30 pages long, but we'll do our best.

Sarah also had a suggestion in the spirit of truth in advertising from the title, I guess it would be. So using the title that we had decided upon, but then "The Study Priorities of the Secretary" rather than "A Report of The Study Priorities of the Secretary's Advisory Committee on Genetics in Health and Society." So it makes it a little clearer in the subtitle that there was a priority-setting process. Is that okay with everyone?

DR. WINN-DEEN: I'm going to try it one more time. Can we get -- Alan?

DR. GUTTMACHER: Again, at the risk of wordsmithing, I think it might be an important concept. I've got a question on page 1 of the executive summary, the second bullet. "Public discussion of the nature of genetic information (conceptualized in the term genetic exceptionalism)." Is that to suggest that genetic information is equivalent to genetic exceptionalism? What is actually conceptualized? I find that unclear and/or misleading.

DR. SHAMANSKI: It was the nature that we're referring to.

DR. GUTTMACHER: The nature of genetic information is --

DR. SHAMANSKI: Is it unique in some ways from other types of information?

DR. GUTTMACHER: Then I think we should be clear, if that's what we mean. "Public discussion of whether or not genetic information is unique" or something like that, because there's certainly much to be discussed about the nature of genetic information beyond the question of its uniqueness. If I'm the only one disturbed by this, then just leave me disturbed.

(Laughter.)

DR. SHAMANSKI: That's fine.

DR. WINN-DEEN: Should we just add the word "unique" in front of "nature"?

DR. McCABE: Why don't we state it "Public discussion of whether genetic information is unique medical information"? That's the real nature of this discussion, as I recall it.

DR. WILLARD: Say "unique personal information." It's not limited simply to medical information.

DR. McCABE: "Unique personal information."

MS. CARR: Also, I think the only reason we were -- the genetic exceptionalism term is not exactly -- I mean, it's sort of esoteric. I think it's what the community knows and uses. So we were trying to not use that term --

DR. GUTTMACHER: Oh, sure. Using both terms is fine, but I just wanted it

clear. Thanks.

DR. McCABE: So the way we have it now is "Public discussion of whether genetic information is unique personal information"?

DR. WINN-DEEN: Deb?

DR. LEONARD: Can we leave "medical" in there? Because it's the medical treatment of the genetic information. I mean it's personal information as well, but can we say "medical and personal information"? Because it's the issues surrounding how you treat that medically and whether it goes in the medical record and things like that.

DR. GUTTMACHER: Isn't it the question of whether genetic information is different from other medical information?

DR. SHAMANSKI: Well, it goes beyond that, because our decision in the end was to look at genetic exceptionalism about each issue as we go. So in terms of education, it's whether we're going to treat the education in genetics differently than we're going to treat others. So the idea of genetic exceptionalism goes I think maybe beyond the information, so maybe we need to work with that wording a bit more.

DR. McCABE: Why don't we include that in the prose as an overarching, that genetic exceptionalism is an overarching --

DR. SHAMANSKI: It is in the list.

DR. McCABE: Okay.

DR. WINN-DEEN: Going once, going twice --

DR. FEETHAM: I appreciate your discussion on the medical. But again, I go along with it's the individual's information. Medical to me is more narrow. It's really health information. It's moving towards electronic health records. I mean, I just think it's conceptually much broader than medical.

DR. WINN-DEEN: So if we said "personal health information," would that capture it?

I think what I'd like to do, unless Ed has other comments --

DR. McCABE: Well, Sarah points out that we need to be accurate also, and genetic information is in fact unique personal information. We are unique genetically.

DR. WINN-DEEN: Personal and health?

DR. LEONARD: Could we say "Public discussion of whether genetic information is different from other personal health information"? Because it's really the difference between. Is there a difference, or should it be treated just like all other personal health information?

DR. McCABE: Any objections to that?

(No response.)

DR. WINN-DEEN: All right. So at this point, I think what I'd like to do is ask staff to make the changes that we've requested, to perhaps send them out to the task force or to Ed and myself.

DR. McCABE: I would suggest a fairly small group to work on this. So why not you and me and Emily, if that's acceptable to the committee, that we will then finalize this document and send it on with a cover letter explaining its nature to the Secretary.

DR. WINN-DEEN: Do we need to get the Secretary's approval before we would post these, or what would we do in terms of public access?

MS. CARR: Well, the process will be that we will send it forward to the Secretary, and once it's received, we can post it on our website. We want to make sure the Secretary has it before we make it public as a final document.

DR. WINN-DEEN: That's probably a good order.

DR. McCABE: Just to point out, that's a term of art, "received by the Secretary." It's not like when we receive a letter. It's when it's been formally received. So it may take a process of weeks to be received by the Secretary, just to make it clear. But that's why I

would like to move forward, because again looking at the calendar, I think it would be good to get it to the Secretary as soon as possible.

DR. WINN-DEEN: Ed, do you want to go ahead and take our break now, or do you want to go ahead and start the next discussion area?

DR. McCABE: Do I have a sense of the committee? Is there anyone that disagrees with this process, then, as outlined to the committee?

(No response.)

DR. McCABE: If I do not hear any disagreement, then we will move forward with that process, make the changes with some dispatch, and send the report on to the Secretary.

Thank you very much. This has been a huge amount of work but I think a very important priority-setting exercise that we went through, and I think it's a good summary of the thinking of this committee and will be useful in a lot of different venues.

Actually, we had the break scheduled -- I think that's a good idea. Why don't we take a break now, Emily, and then we'll have the public comment after the break.

Debra?

DR. LEONARD: Can I ask a question? The education resolution, was that revised and will we see that this afternoon during the working session at lunch? Just for information.

DR. McCABE: Why don't we take the break now? Yes, I think we will have time, but let's keep this to a 15-minute break.

MS. CARR: Yes, it's been revised, and I think the plan was to bring it back to the committee to show the revisions at some point during the three-hour open session, I think, and then there may also be some additional discussion of the coverage and reimbursement report. Is that correct?

DR. McCABE: Yes.

Let's take a 15-minute break, and we will resume at about 10:50.

(Recess.)