Development of Final Recommendations (continued)
Facilitators: Reed V. Tuckson, M.D. and Andrea Ferreira-Gonzalez, Ph.D.

DR. TEUTSCH: Folks, let's reconvene. Cathy, did you have a response to a query from Reed? I'm not even sure which query it is.

DR. FOMOUS: Yes, I do. Reed asked how many of our recommendations are calling for public-private partnerships or stakeholder groups as part of the recommendation. So in direct answer to Reed's question, there are three. There are our revised Recommendations 3 and 4 in Chapter 4 that call for these multi-stakeholder groups. They theoretically could be the same group of people. Then Recommendation 1 in Chapter 5, asking for a multi-stakeholder group to look at clinical utility issues.

Now, in addition, there are a couple of recommendations that suggest that relevant agencies might want to engage a group of stakeholders just for additional information. That kind of connoted less permanence.

DR. TUCKSON: Thank you. Let's keep track of that as the conversation goes forward. My suggestion is that when we get to the end, before we finalize all of our vote, we take one more look at exactly how much bucks we spent and how many committees we convened and see whether that affects any decisions.

DR. WILLIAMS: May I ask a question related to that? Is there a threshold over which we say that this is not important and therefore we shouldn't make the recommendation?

DR. TUCKSON: I don't know. I have no idea. What it may do, also, is say certain things may need to be combined. I don't know. I have no idea what it is going to look like.

DR. FERREIRA-GONZALEZ: Why don't we keep plowing through and then we will get to that point.

DR. TEUTSCH: Yes, I'm looking forward to that moment. So, Andrea and Reed, walk us through the last of these.

DR. FERREIRA-GONZALEZ: Recommendation 3 for the registry, we are going to add the wording of "least burdensome."

So Recommendation 5 requests enforcement of existing regulations. We revised Part A to include that laboratories without CLIA certificates cannot be reimbursed by Medicare and Medicaid but these restrictions have no consequences for laboratories that perform direct-to-consumer testing.

We did not make any changes to Part B of the recommendation.

Do you have any questions about this recommendation?

[No response.]

DR. FERREIRA-GONZALEZ: That is refreshing. Do you have any edits?

DR. EVANS: Yes, I'm sorry.

DR. FERREIRA-GONZALEZ: Jim.

[Laughter.]

DR. EVANS: I'm just wondering if we want to make it a little bit stronger. It seems awfully tepid, saying they should explore mechanisms, develop new authorities and resources. Say they should find ways to close that loophole.

DR. FERREIRA-GONZALEZ: So, what language would you recommend? If you are going to speak up, you have to come up with some.

DR. EVANS: How about just forget "explore mechanisms and seek." Just "should develop the authority" or "implement its authority in order to effectively enforce."

DR. FERREIRA-GONZALEZ: But they might not have the authority, so they need to seek.

DR. EVANS: "Should develop the authority" or "attain the authority" or just something a little stronger.

DR. FERREIRA-GONZALEZ: Judy is not here, so.

DR. EVANS: "Should develop the authority." How about "develop the authority"?

DR. TEUTSCH: Jeff, do you happen to know what the authority is?

DR. ROCHE: No, I don't.

DR. TEUTSCH: How about just saying "should strengthen its enforcement efforts against laboratories."

DR. FERREIRA-GONZALEZ: Whatever they have to do, seek authority or not, then they will do it.

DR. EVANS: There you go.

DR. FERREIRA-GONZALEZ: Repeat that again, Steve?

DR. TEUTSCH: "HHS should strengthen its enforcement efforts against laboratories." You just delete everything from "explore" down to "to."

DR. FERREIRA-GONZALEZ: There we go. Very good. Can we move to the next recommendation?

DR. TUCKSON: I just want to make sure. Until they step up, does this leave a hole?

DR. FERREIRA-GONZALEZ: Yes.

DR. TEUTSCH: Yes.

DR. TUCKSON: I just want to be even more explicit. We need to make sure that everybody who reads this understands that you have to close the hole. This is like saying they should step up, we hope that. I think you have to say the explicit intent here is to close the hole.

DR. EVANS: "Should close this gap."

DR. TUCKSON: And that the only way you can do that, we are saying here, is --

DR. FERREIRA-GONZALEZ: But, isn't the first sentence saying that?

DR. TUCKSON: It says "Further efforts are needed to prevent laboratories." I guess that says close the hole.

DR. FERREIRA-GONZALEZ: Actually, it should be CMS.

DR. TUCKSON: The question is, you can't impose them on uncertified laboratories. You can't impose it on an uncertified lab. Remind me what the uncertified lab is, again?

DR. FERREIRA-GONZALEZ: A laboratory that doesn't have a CLIA certificate. Today the problem is that when they go to inspect the laboratory or they have come upon a laboratory and they don't have a CLIA certificate, they cannot close down the laboratory.

DR. TUCKSON: That is kind of crazy. It doesn't make sense to the average person. In other words, I inspect you, you don't qualify, I can't inspect you. That can't be it. That is what it sounds like. Because you are so bad, I can't do anything to you.

DR. TEUTSCH: They have to be referred to the inspector general for enforcement, is the problem.

DR. FERREIRA-GONZALEZ: Yes. Then the inspector general at that point intervenes. So we are asking them to have something direct. If they come across a laboratory and they don't have a CLIA certificate and they are doing clinical laboratory testing, that they can do something to that laboratory right there.

DR. TUCKSON: That is what this is saying, that they can now stop you.

DR. FERREIRA-GONZALEZ: Without having to go through another.

DR. TUCKSON: Enforcement action. So "CMS should strengthen its enforcement action against" -- no. "CMS should have enforcement action against."

DR. TEUTSCH: "Should obtain and strengthen," is that the issue?

DR. KHOURY: A quick question. Are we conflating DTC with no CLIA certification?

DR. FERREIRA-GONZALEZ: No, no, no. We are talking about laboratories --

DR. KHOURY: The section in green. If you read it again, "Labs without CLIA certification cannot be reimbursed, but this restriction has no consequences." Can you explain that again?

DR. FOMOUS: The first part of this sentence would have consequence for labs that are actually performing clinical testing that is related to health care or tying into results that patients need. There would be a consequence for that. But for a lot of these DTC companies that we are concerned about, they don't care about reimbursement because the consumer is paying them directly.

DR. KHOURY: If a DTC company has CLIA certification, what will happen?

DR. FERREIRA-GONZALEZ: If a DTC laboratory has CLIA certification? Nothing. They have CLIA certification. They are okay. If it is a laboratory, DTC or not, that doesn't have a CLIA certificate --

DR. KHOURY: Some of them do.

DR. FERREIRA-GONZALEZ: Even if you do have a CLIA certificate, then you are not covered in here.

DR. WILLIAMS: We shouldn't confuse the two issues. We have addressed in other recommendations the fact that some of the tests that are being done in CLIA-certified labs don't have validity and utility. So we are trying to address that in a different area.

This is specifically addressing those laboratories that do not have CLIA certification that are performing tests that we consider to be health-related and should fall under CLIA. The only enforcement ability that CMS currently has is to not reimburse. We are seeking enforcement ability beyond that, which is to say you must cease and desist.

DR. FERREIRA-GONZALEZ: Steve.

DR. GUTMAN: I'm sorry that Judy is not here, but I'm sure that is wrong. It is illegal in this country to offer a lab result for a medical purpose on any person in a non-CLIA-certified lab. I don't know what her tools are.

DR. FERREIRA-GONZALEZ: We understand that it is illegal.

DR. GUTMAN: Now, they may not be enforcing them as enthusiastically as you would like, but

DR. FERREIRA-GONZALEZ: But it is not only enforcing, it is the way that she currently has the tools to enforce are going through a different mechanism. There is nothing very direct right there to do something about that.

DR. GUTMAN: I would argue it is very direct. I don't want to pick any names, but if you look at the companies that were described at the Smith hearing, many of them were non-CLIA-conformant before. They are all CLIA-conformant now. I think she has more tools than you understand.

DR. FERREIRA-GONZALEZ: She was okay with this recommendation.

DR. GUTMAN: I'm sorry she is not [here.]

DR. TEUTSCH: Judy, are you on the phone, by any chance?
[No response.]

DR. GUTMAN: I think we should call her and ask her about this. Unless she is wanting encouragement to use those tools more liberally, I'm certain she has them.

DR. FERREIRA-GONZALEZ: She has seen these recommendations and we haven't heard anything specific from her.

DR. GUTMAN: She is the CLIA expert, not me, so I will shut up.

MS. CARR: She has been on the phone on and off this morning, and I think she is en route back to her office. I think she will be on shortly. I just Emailed her to see. We will try to get her. But she did review this recommendation. She actually provided us input that helped us clarify the role the inspector general has.

DR. FERREIRA-GONZALEZ: Yes, that is how we learned about the inspector general, that route, from her.

DR. GUTMAN: Again, she is the expert, I'm not. But I do know she has tools. So her motive must have been perhaps giving her more --

DR. FERREIRA-GONZALEZ: It is more direct.

DR. FOMOUS: So, did we want to change the wording here?

DR. FERREIRA-GONZALEZ: Yes. Reed, what do you want to change here? Is Reed there?

DR. TUCKSON: I don't like this "further," but anyway, that is wordsmithing. First of all, what I would like you to consider is taking "further efforts are needed" and saying "to prevent laboratories from performing genetic tests without appropriate CLIA certification," here is what you should do. I think that is what it needs to be. You just go straight to the heart.

So, "To prevent laboratories from performing genetic tests without appropriate CLIA certification, the Committee makes the following recommendations." I guess that is repeating, maybe, the preamble. But anyway, without getting into that.

So then you need to say that "The CLIA program has an array of enforcement actions available but those actions cannot be imposed."

DR. TEUTSCH: Judy, are you on the phone yet?

MS. YOST: Hello. I just got to my office.

DR. TEUTSCH: Oh, Judy, good. You are very timely. We are looking at Recommendation 5 in Chapter 4. Steve raised the issue regarding your comment on the need for CMS to secure additional enforcement activities against labs which are not CLIA-certified. We believe you reviewed this recommendation, but are there issues here that you need to raise about whether you already have those authorities?

DR. TUCKSON: Judy, this is Reed. I'm the one that is struggling with it, and Steve has introduced it well. What we are specifically trying to get at here is that the impression that we are

left with from the original recommendation is that you do not have the authority to specifically regulate non-CLIA-certified labs and, as a result, the only power you have is to go to the inspector general to have that person's office fill the gap.

We are trying to understand that because it seems to us that it doesn't make any sense, that what we ought to basically say is you should have the authority to regulate the labs, whether they are CLIA-certified or not.

MS. YOST: The legal answer is yes, that is essentially what the regulations provide for. They provide that if we have to take an action against a laboratory that does not have a certificate, in that case it would have to be referred to the OIG. However, we have put into place mechanisms for those circumstances when we come across them and have dealt with them rather successfully.

That doesn't say that we probably maybe need some more, but we have been able to deal with them fairly successfully by sending them a rather unfriendly letter that says that they must cease and desist their testing because they are operating outside of federal law. In most cases, that is rather effective. We do not allow them to initiate testing again unless they apply and their application is approved to begin testing again. That has worked.

So it is not a total black-and-white answer.

DR. TUCKSON: That is helpful. That is very helpful. In terms of moving this forward, if we were to just take out the ambiguity of all of this and the jerry-rigging of it and scaring people, what if we were to simply make logical sense and recommend that CMS ought to have the authority within its body unambiguously to perform this function. Is that something that has to be done through statutory change?

MS. YOST: I would assume you could do it through regulation.

DR. TUCKSON: Through regulation?

MS. YOST: We can throw it in the PT regulation.

DR. TUCKSON: I know you have the awkward position of not being able to actually write it, but we are asking you specifically so that we don't have confusion. We would recommend that the Secretary take steps to seek -- who writes the regulation?

MS. YOST: CMS and CDC write the regulations. For enforcement, CMS would do it.

DR. TUCKSON: So, can you write this sentence for me? If we said that the Committee recommends that the Secretary of Health cause the following agencies to write the regulations that will permit CMS to regulate non-CLIA-certified labs.

DR. TEUTSCH: "Should secure the regulatory authority."

DR. TUCKSON: From where? "Secure the regulatory authority from"? Congress?

MS. CARR: It doesn't need statutory.

DR. FERREIRA-GONZALEZ: It is not statutory.

DR. TUCKSON: So, where do they get it?

MS. CARR: Themselves.

DR. WILLIAMS: They write a Federal Register announcement and get comments.

DR. TUCKSON: So they get it from themselves?

DR. WILLIAMS: Right. So they should assume regulatory authority.

DR. TUCKSON: So there is the language. Judy, would you be okay with that?

MS. YOST: Yes, that's fine. That is why I didn't say anything else with what was there, because I had a feeling that that would happen anyway. But I did want to explain that we do have some and it has worked. In fact, for the illustrious folks we had on the phone yesterday, the laboratory that they are currently using just underwent that process successfully.

DR. TEUTSCH: It should say that "CMS should assume the regulatory authority to allow it to take enforcement actions against laboratories that perform."

Any other comments on this?

[No response.]

DR. FERREIRA-GONZALEZ: So we have the language. Do you have it, Cathy?

[Pause.]

DR. TEUTSCH: "Regulatory authority to allow it to take enforcement actions against laboratories."

DR. FERREIRA-GONZALEZ: So we have to put "to allow it to take." No, don't take the "enforcement."

[Pause.]

DR. TEUTSCH: It already has authority.

DR. FERREIRA-GONZALEZ: They already have authority.

DR. TEUTSCH: What are you asking it to do is be able to enforce those actions directly itself as opposed to referring it to a third party.

DR. FERREIRA-GONZALEZ: Hold on, hold on. "Should exercise its regulatory authority to allow."

[Pause.]

MS. YOST: This is Judy again. I wanted to mention, too, because I don't know where you are in the process of this because I kept getting disconnected from the telephone. I don't know if you

had any further discussion about the DTC labs, which is kind of related here. I know some of the concerns about those labs --

DR. FERREIRA-GONZALEZ: It is coming up next, Judy.

DR. TEUTSCH: It is the next item.

MS. YOST: Oh, okay. I will be quiet.

DR. TEUTSCH: Anything else here?

DR. FERREIRA-GONZALEZ: Yes. Anything else that we want to add to Recommendation No. 5?

[No response.]

DR. FERREIRA-GONZALEZ: Let's move to Recommendation 6. Now, remember we had some issues in Chapter 6, Recommendation 5 on how we were going to tie it up with this one. So let's get back to Recommendation 6.

Recommendation 6 calls for the expanding CLIA regulations for CMS's statutory authority through CLIA to encompass certain direct-to-consumer tests that appear to fall outside CLIA's scope. We revised this recommendation to include FDA's authority and regulatory process.

Do you have any questions about this recommendation? Judy, you made some comment about this. Is there anything you want to add to this recommendation?

MS. YOST: I actually was just going to talk about the DTC labs because I know there is a lot of concern about the ones who are advertising over the Internet and whether or not they are CLIA-certified.

I just wanted to let the folks know that we have taken it upon ourselves and we are collaborating with both CDC and FDA that when a laboratory like this is identified that we will follow up to investigate what, if any, type of testing the laboratory is performing. For those that are within the current scope of CLIA, that they do obtain a CLIA certificate.

Those efforts have been rather successful, to the point where we currently have 64 laboratories identified that we have reviewed and not only do we just do an initial investigation, we do continuous follow-up until we are satisfied that they are not only enrolled but in compliance.

So I just thought you should know that we haven't been sitting around.

DR. TEUTSCH: That is very helpful, Judy. This is really talking about expanding the scope.

MS. YOST: Right. I realize that. But I did want to throw that in there because I think that people think we are just ignoring that, and we are not. We are very well aware of it and we are going to try and set up something more formal to accommodate that.

With regard to scope, we have had discussions with our attorneys and clearly, right now at least, in order to come under the purview of CLIA an entity would have to meet the current statutory definition of a laboratory. So that is what our limitations are currently.

DR. FERREIRA-GONZALEZ: Any comments? Marc.

DR. WILLIAMS: The only point I would make is that it seems now, as we look at this, that Recommendation 5-B would fit better as part of this recommendation because this deals with issues of claims which will be directly impacting the determination of health-related. So I just think moving 5-B and incorporating it No. 6 would make it clear.

DR. FERREIRA-GONZALEZ: Yes, I think it is a good idea.

DR. TEUTSCH: The one before isn't limited just to these DTC, whereas Recommendation 6 is.

DR. WILLIAMS: All right. I withdraw my [suggestion.]

DR. FERREIRA-GONZALEZ: We don't have any comments. Any edits to this one?

[No response.]

DR. TEUTSCH: We have been through them all once now, correct?

DR. FERREIRA-GONZALEZ: Yes, yes. Well, we still have one more. Sorry to tell you. We have an overarching recommendation.

DR. TEUTSCH: Oh, correct.

DR. FERREIRA-GONZALEZ: So we have an overarching recommendation that outlines steps to enhance interagency coordination for oversight activities. Do we have any comments? Yes.

DR. WISE: I'm concerned that this is pretty weak. It doesn't speak to the concerns that Reed has been raising straight through all these conversations.

Right now it reads sort of as an insider's report, which is totally understandable given the technical complexities and what has to be dealt with. But genetic testing is also an issue of great public concern and there has to be a framing, I think.

We may want to take advantage of the overarching recommendations to help frame the public presence of this report in language to say genetic testing is expanding greatly, however oversight of genetic testing currently is inadequate. Therefore, the issues we have identified looking at the gaps, there are jurisdictional problems that would relate to coordination, there are authority issues, there is quality control, and then there is dissemination of appropriate innovation to the people who need it. All the gaps fit into those categories.

So, is there a way to use either the overarching recommendations or something up front that would help frame the public presence of this report in a way that translates the technical conversations and the technical language that is in there into something that makes this more accessible and controls the public presence more than the way it is written now.

DR. FERREIRA-GONZALEZ: So, what would you recommend?

DR. WISE: If people are happy with the idea that in fact the report needs to do this, to try to accomplish this, then my suggestion would be to not elevate what we need just to get along better

and then elevate the other themes that have come through, one including better coordination, but the issue of filling major gaps in regulatory control.

There are sentences in here, but to elevate, to use this mechanism to elevate the critical positions, the legitimacy, the justification for this report in a way that is accessible. If this isn't the best way, then I'm happy to try to do it a different way.

DR. TEUTSCH: Paul, you can see it in the executive summary. Between the header, "Recommendations," there is some text that is sort of the preamble to all of them. What I'm hearing you say is you are not thinking just to this, you are speaking to all this set of recommendations.

I wonder if we can't craft some language that will sit in that space. These are the issues we have found and the recommendations below speak to them.

DR. TUCKSON: Steve, one thing I would speak to is I do like where Paul is going and your comment. The word that is missing in all of this is the word "accountability."

To me, I think that that is the overarching recommendation. It is saying that you take all the things Paul said and, at the end of the day, the recommendation is that the Secretary must use all of his or her power in terms of the agencies reporting to them and is accountable for protecting the public in this regard. Then the recommendations that come down the pike start to get more specific about those things, but the word "accountability" is where I see the overarching.

DR. FERREIRA-GONZALEZ: That is a different issue. the issue that we have here, too, is that there might be different activities happening out of the Secretary's office that might be duplicative or somewhat of an overlap. That is what we are trying to get here, that the Secretary needed to have a better coordination of the different activities that are currently happening under his office.

DR. WISE: The issue is, should that be the overarching recommendation, the one that is elevated above all others. There has not been a report about the federal government that has not included this issue, ever. My concern is, given the importance of this issue, that we need a framing recommendation that truly gets to the heart of why this is so important.

I agree; I think the accountability may be the overarching recommendation and that the preamble and the executive summary or that text that is there now can be reworked to be more focused, more clear, and to state that action steps are going to be required to optimize the benefits of this new technology but also to prevent the harm that also is potential outcomes.

To take the gap analysis that is very technical, and that is required to be technical, and to reframe it as the three or four big ticket arenas of action that are going to be required. It doesn't have to be the overarching recommendation. I like Reed's. But to have something right up front that frames this report and that sets the foundation for the interpretation of the technical language that ensues.

DR. FERREIRA-GONZALEZ: Muin.

DR. KHOURY: I want to second what Paul was saying here. I think that you can beef up the preamble and all the background, but given how the Committee has worked so hard to identify this monster here plus all the gaps, et cetera, the recommendation to the Secretary to say "Take steps to enhance interagency coordination" is rather weak.

I have been in other committees where, at the end, an interagency working group was created, for example. Even that was weak because it got disintegrated over time.

But you have to tell something to the Secretary that is a little bit more substantive than just taking steps to enhance interagency coordination. That is what they are doing all the time. What kind of steps do you want them to take.

One approach is to create a working group that would oversee the implementation of the recommendations or do all this in part of the Personalized Healthcare Initiative, which genetics fits nicely under.

It will obviously be left to the next administration to implement, but you need to send a stronger signal than just "take steps to enhance interagency coordination." Just different words.

DR. FERREIRA-GONZALEZ: Joseph and Marc.

DR. TELFAIR: This is just a question related to this. As a public health person, one thing that we look at a lot is who are our target populations and who we deal with. I think "the interagency coordination for the purpose of reporting to," and then whoever we are deciding who the target population needs to be accountable to, be it providers, be it the public, or whatever, as a strong statement coming out of an overarching recommendation.

If you look at everything else that has been said over all these recommendations, you have parceled out who these target populations are and who should be accountable or moving towards.

I would just say it should start off with an outcome statement right off the bat that is pretty strongly stated and includes what it is you are trying to do. The Committee itself can look at what they are recommending, but it should be for the purpose or for the expectation that whoever, the public, the providers, the other persons who are constituents, will be able to enact or be able to be involved with this group.

Something in that neck of the woods seems to make sense. I think you start off with a very strong outcome statement to move forward with that.

DR. FERREIRA-GONZALEZ: Marc.

DR. WILLIAMS: Personally, I think if we could capture what Reed said and substitute that as an overarching recommendation for what we have here. As I read through this again, most of it is represented in the other sub-recommendations.

DR. FERREIRA-GONZALEZ: This was also speaking to some issues that we started identifying, that within the Office of the Secretary there are different agencies or groups that are working on similar issues at the same time.

DR. WILLIAMS: I recognize that, but I think we reflect that in all of our recommendations because we have the same alphabet soup that is appearing in all of them. I think, to be very clear, the issue is protection of the public and gaps must be closed. That should be the overarching recommendation, and the rest of it is going to fall out.

DR. FERREIRA-GONZALEZ: Jim.

DR. EVANS: I agree exactly with what Paul and Marc have said. I think that this is all about the fact that gaps exist, harm could result from those gaps, the gaps have to be closed. The rest of the recommendations all pertain to ways that we recommend to close those gaps.

But I think an overt statement at the start along those lines would strengthen the report immensely.

DR. FERREIRA-GONZALEZ: Kevin and then Reed.

DR. FITZGERALD: If we just take what we have at the beginning there, which I think is still a nice little setup, there is this complex oversight system, many dedicated people. Hold on, wait a minute. Then say, "Nonetheless, the Committee also found significant gaps in the system that could and do lead to harm. Therefore, the Committee recommends," or we can put it that way. "The Committee states that the Secretary of Health and Human Services should take accountability for addressing these issues. We have, in the following, put forward some specific recommendations," but ultimately it all ends up in the Secretary's lap. Period.

DR. FERREIRA-GONZALEZ: Julio?

DR. LICINO: What is the actual harm that has occurred as a result of the current system? If there is, we should document it. If there isn't, we should say "potential." But I'm not aware myself of actual harm to anybody due to the current system. I may be missing something.

DR. TUCKSON: Folks may want to start thinking in their mind whether the word is "potential" or "actual" or do we know enough to know. Because you don't know what you don't know. I think the stuff that we have been hearing around the direct-to-consumer stuff where you just have no idea what pop-ups are going where; you just don't know. It may be that we have some documentation that we want to bring forward or we just want to say "potential."

Be that as it may, I think I liked everything that Kevin said to advance the ball. The only modification, and I think it is not even really much difference, it is very much informed by Muin showing the map. It sounded like I was hearing Kevin say the Secretary is accountable for getting these recommendations done.

I think what I'm saying is, look, the Committee has done its very best by providing a set of recommendations. It is almost to Paul Wise's point. We have done the best we can under the time constraints that we had. And I don't want to diminish our recommendations by doing that.

We have given a variety of recommendations which we think move it forward. At the end of the day, this is complicated. The Secretary ultimately is accountable for making this complex puzzle make sense to protect the public. At the end of the day, that is where this falls. People should not be hoping, praying, trusting that we have it all. At the end of the day, Mr. Secretary, you have a bunch of people who are very smart and very dedicated. You have to make this happen.

DR. WILLIAMS: The point I would just make about the harms is that the text of the report clearly identifies those harms for which there is literature, support, and clearly identifies those harms that are potential or plausible but for which there is no documentation. I don't think we need to revisit that.

DR. FERREIRA-GONZALEZ: No. So we need to start working on the language.

DR. TUCKSON: One last question I have before Andrea brings us to whatever our summary is. I think we heard that we didn't add any more committees and any more money, so I think we just need to take one more look at that summary again.

But also, did we resolve Mara's point? I'm not sure whether we answered what is in and what is out as the definition of a genetic test. Did we resolve that?

DR. FERREIRA-GONZALEZ: Don't go there.

DR. TUCKSON: Don't go there? I can leave it alone? It's too late? We are all right? All right. There is a strong consensus to not raise that. I just wanted to make sure we weren't forgetting something.

So, give us the numbers again. How many committees did we create?

DR. FERREIRA-GONZALEZ: Have we finished with the overarching recommendations? I don't see him writing.

DR. TUCKSON: So, what was the amount of money and the committees again?

DR. TEUTSCH: Three committees.

DR. TUCKSON: Three committees. What were those again?

DR. TEUTSCH: Registry, utility, and --

DR. FOMOUS: FDA review and registry, which could be one and the same, and then one for utility.

DR. TUCKSON: That is why I wanted you to slow down. You said they could be one and the same. Let's go back and understand that.

DR. FOMOUS: Registry and FDA review.

DR. TUCKSON: So registry and FDA could be one and the same. Now, this may be beyond your neuronal capacity at this point, since we have dangling participles here, Marc. But, is anybody prepared to think about can we put those two together and is that going to be harmful?

I really, obviously, am trying to get us to where you don't have three, if you can get two, because it just gets to be a god-awful nightmare trying to administer this stuff.

PARTICIPANT: To me that seems to be micro managing. We don't need to specifically articulate that they have to be separate committees, and I think we could leave that to the Secretary to decide what is the best way to do that.

DR. TUCKSON: I'm suggesting you take what Marc said and capture it in the letter of transmittal from the Committee. What I don't want to have happen is a loyal staffer to the Secretary walks in and says, "Secretary, I want to brief you on the Committee's report. They are asking you to create a massive new government infrastructure, three committees with 50 people on them. Public-private people from all over the world have to be convened twice. The costs for

travel are going to be, blah, blah, blah. Somebody has to staff it," and the Secretary and these people are out of their minds.

So if you can get away with limiting this to two, that would be terrific. But if you are saying that is micro managing, then, Marc, maybe the sense of the Committee is, Mr. Secretary, we think that you have discretion on how you administer this and we are sensitive to the cost effectiveness of what we are proposing.

DR. FERREIRA-GONZALEZ: Look at the map and the gaps. It might not be feasible to have a single group looking at all this.

But we still have to go back to Chapter 6, Recommendation 5.

DR. TEUTSCH: Are we done with this?

DR. FERREIRA-GONZALEZ: No, they are wordsmithing.

DR. WILLIAMS: Actually, Andrea, for Chapter 6, Recommendation 5, I believe that as we have rewritten Chapter 4, 5-B and 6, that what was left in Recommendation 5 in Chapter 6 is now completely redundant. I think it is captured in Chapter 4, Recommendations 5-B and 6. So I think we should just take it out.

Now, that is not my decision to make, but that is the conclusion that I come to.

DR. FERREIRA-GONZALEZ: Why don't we take it out as we go through it again.

DR. WILLIAMS: That is what I'm saying. Let's not visit it now.

[Pause.]

DR. WILLIAMS: So, start with "The Committee."

DR. FOMOUS: We are not going to use any of this?

DR. WILLIAMS: Yes, we will be, but it is moving around too much. Ready? "The Committee found significant gaps in the U.S. system of oversight of genetic testing that could and do lead to harms." Julio, we can talk about this, but we do have data that say there are harms.

"The Committee formulated a number of recommendations that, if implemented and sufficiently supported, could close these gaps. The Secretary of HHS must take responsibility for closing these gaps and fostering the public health."

That's it. They wanted something simple, direct, and overarching.

"Public's health"? That would be all right.

[Pause.]

DR. FERREIRA-GONZALEZ: I'm still confused about this. That is why we are giving him the report. He must do.

MR. MILLER: I would just change that second --

DR. FERREIRA-GONZALEZ: Remember we are an advisory group.

DR. WILLIAMS: And we are advising him.

DR. FERREIRA-GONZALEZ: Yes, all these recommendations.

MR. MILLER: I would just say "The Secretary of HHS is responsible for closing these gaps." Because anybody can say "I'm taking responsibility for everything." Secretary Leavitt, close these gaps.

[Laughter.]

DR. TEUTSCH: How about just saying "should close these gaps"?

MR. MILLER: No. He is responsible for closing the gaps.

DR. LICINO: Can't we just say "closing these gaps" --

DR. FERREIRA-GONZALEZ: Julio, turn over your speaker. We are taping.

DR. LICINO: Can't we just say that closing these gaps is in the public health interest"?

PARTICIPANTS: No.

DR. FOMOUS: Can we just alter the end of it a little bit to say --

MR. MILLER: Please.

[Laughter.]

DR. FOMOUS: Please, because I think it will make it even stronger -- "is responsible for closing these gaps to foster" or "to optimize the public's health"?

DR. FITZGERALD: I guess the idea there was one can talk about closing the gaps but there is also the possibility of going beyond just closing gaps. One can, once the gaps are closed, still continue to work to foster the good that can come from these technologies.

So if you limit it to just closing the gaps, then the work is done. But I think some of what we have been talking about is the fact that there is more to do than just closing gaps.

DR. FROHBOESE: That gets at the main point, that there are gaps, here is what we recommend, and not only is it just about the gaps but the big picture is the public's health. That is the big picture.

DR. FERREIRA-GONZALEZ: This is an overarching recommendation, but we still are going to have another recommendation as part of this one to talk about the coordination?

DR. TEUTSCH: No.

DR. LICINO: Just one thing. I'm a little troubled by this. He convenes an advisory committee who says "You have to do this." I would say that it is our advice to the Secretary of HHS that these gaps be closed to foster the public's health," because that is what we are supposed to do, give advice. We are not supposed to tell him, "It is your job to do this." It is very strange. I feel kind of strange about this.

MR. MILLER: I would say that he has convened this panel of experts to look at a particular issue. I don't think we are saying anything particularly radical here by saying that the HHS Secretary is responsible for closing these gaps or responsible for his agency. We are laying it on his desk to say here is what we think you need to do.

DR. TELFAIR: There are two things. First of all, it is an expectation that it is his responsibility. It is an expectation. If we read through pretty much all the comments, particularly the public comments, there is an expectation listed here.

Second of all, by enforcing an after-the-outcomes effort, which is what we are talking about when you look at the "and fostering the public's health," is adding teeth to a lot of the things that are going to come after it with this set of recommendations. That is pretty clear what we are asking to be done.

DR. FITZGERALD: If we are worried, we could always say, "And if you have any questions about this, see Reed Tuckson."

DR. FERREIRA-GONZALEZ: Martin.

MR. DANNENFELSER: How about a sentence like, replacing that last sentence, "This action is consistent with the Secretary's responsibility for fostering the public's health"?

MR. MILLER: With all due respect, let's just say what we mean. The Secretary has prompted this Committee to go through extraordinary efforts to get him a document because he wants something before he leaves. That is where we are. We are just saying, you asked us a series of very important questions, we found some very significant problems, we came up with our best efforts to think about it. It is now your responsibility to do something.

I don't think that that is overreaching. I don't think it is impolite. I just think it is appropriate.

DR. FERREIRA-GONZALEZ: We are done.

DR. TEUTSCH: I want to go back to one thing Marc said. I'm okay with removing the one recommendation, but we need to make sure that in Chapter 4 where we talked about the oversight of marketing and other such things that we are explicit that that should include the DTC and tests as well.

DR. WILLIAMS: Yes. That just is report modification.

DR. TEUTSCH: Right. That is the editorial kind of thing that we can take care of.

DR. FERREIRA-GONZALEZ: Yes, we have been through everything. Do we want to go through all the recommendations?

DR. TEUTSCH: Why don't we do this. Why don't we read through the recommendations one time now. Are there other things that are missing that are not here? This is our last chance to add new recommendations.

[No response.]

DR. TEUTSCH: Do you want to go ahead and vote now and walk through it? Sarah, what is your advice on how we proceed?

MS. CARR: If the Committee will consider it, I think you ought to try come to a consensus now on the body of your work here. Final recommendations and that the draft report is in spirit ready to be sent to the Secretary after you have an opportunity by February 20th to provide some additional comments. No additional input on the recommendations.

DR. TEUTSCH: So, do we have them so that we can read through all of the recommendations?

DR. FERREIRA-GONZALEZ: Just a reminder on the voting. Questions to consider in voting for the recommendations. Are these recommendations the optimal way to address the opportunities and challenges identified in the report, and are these the recommendations that the secretary of SACGHS should make to the Secretary. Those two we have to keep in mind.

Now we are going to go in order, actually. Just let's go. Just go through it.

Chapter 6, Recommendation 1.

DR. TEUTSCH: Let's go through it in the right order this time.

[Pause.]

DR. WILLIAMS: Mr. Chair? Dr. Chair? It seems to make sense at this point, since we really have significant organizational things, that we should take a lunch break, reorder the recommendations so that we can go through them in order.

DR. FERREIRA-GONZALEZ: What time is Reed leaving? Reed is going to be leaving at one o'clock.

MS. CARR: Do you feel like you can't just go through them?

DR. WILLIAMS: I'm just looking at recommendations that are right now all over the place. It just seems to me we will lose a lot of time trying to find which recommendation is where and then putting the next one up. But I think we have to go through them in order.

DR. TEUTSCH: Cathy, where are you? How easy is it going to be to walk through these from what you have there?

DR. FOMOUS: If we do them backwards like we reviewed them initially, we can do them very rapidly. If we go Nos. 6 through 4.

DR. TEUTSCH: No, we need to do them in the right order.

DR. FERREIRA-GONZALEZ: We need to do them in order.

DR. FOMOUS: It will be fine. I mean, there will be a slight pause as we go from chapter to chapter, but that is all.

DR. TEUTSCH: Let's try and walk through them one at a time.

DR. FOMOUS: I would like to recommend, though, that we reserve the overarching recommendation until the end so that we review all the recommendations and make sure that one really captures them.

DR. TEUTSCH: That's fine. You can read it now and we can revisit it at the end if we need to. So, why don't you go ahead.

DR. FERREIRA-GONZALEZ: Chapter 4, Recommendation 1.

DR. TEUTSCH: Are you going to read them to us?

DR. FERREIRA-GONZALEZ: Do you want me to read them? Okay.

"For a number of years CMS has been planning to address gaps in the oversight of laboratories that conduct genetic tests with the addition of a genetic testing specialty under CLIA. Recently, CMS changed directions and is now addressing these gaps with a multifaceted action plan. SACGHS considered CMS rationale and reviewed the agency's action plan. SACGHS carefully considered the recommendations of prior groups as well as the perspective of the stakeholders who support the specialty.

"In the end, the Committee came to the conclusion that identified gaps can be addressed without the creation of a genetic testing specialty. SACGHS proposes the following recommendations to support and/or augment the CMS action plan.

"Recommendation 1-A. Currently, CLIA requires all non-waived tests to undergo some form of performance assessment, but only 83 specific analytes, none of which are genetic tests per se, are required to undergo the type of assessment called proficiency testing. PT is currently considered to be the most rigorous form of performance assessment.

"In principle, genetic tests and all other non-waived laboratory tests should be required to undergo PT. However, such a goal cannot be achieved immediately. Consequently, the following actions should be taken.

"CMS should require PT for all non-waived laboratory tests for which PT products are available. For tests without PT products, laboratories must use alternative assessment methods, as required under CLIA regulations.

"In order to promote the development of new PT products and facilitate performance assessment efforts, HHS should fund studies of the effectiveness of other types of performance assessment methods to determine whether they are as robust as PT, and support innovations in the way PT is performed, such as through methodology-based processes.

"CMS should consult or contract with experts in the field to train inspectors of genetic testing laboratories. Training by such experts will enhance the inspectors' understanding of the technologies, processes, and procedures utilized by genetic testing laboratories and equip them to assess compliance with CLIA requirements.

"In addition, CMS should identify and evaluate innovative alternative mechanisms to inspect genetic testing laboratories.

"As recommended in the 2006 Government Accountability Office Report on Clinical Laboratory Quality, CMS should use revenues generated by the CLIA program to hire sufficient staff to fulfill CLIA's statutory responsibilities. The program should be exempted from any hiring constraints imposed by or on the agency.

"Recommendation No. 2. Currently, there are gaps in the extent to which analytical validity and clinical validity data can be generated and evaluated for genetic tests. To address these gaps, SACGHS recommends supporting public resources for genetic testing through the following actions.

"In consultation with relevant agencies, HHS should assure funding for development and characterization of reference methods, materials, and samples; for example, positive and negative controls and samples from different ethnic and geographic populations for assay, analyte, and platform validation, quality control, performance assessment, and standardization.

"HHS should assure funding for the development of a mechanism to establish and support a laboratory-oriented consortium to provide a forum for sharing information regarding method validation, quality control, and performance issues.

"HHS agencies, including NIH and CDC, should continue to work with public and private partners to support, develop, and enhance public reference databases to enable more effective and efficient collection of mutations and polymorphisms data and expand clinical reference sequence databases, and provide summary data on gene disease associations to inform clinical validity assessments, e.g. RefSeqGene or HuGENet.

"Such initiatives should be structured to encourage robust participation, for example, and may need to consider mechanisms for anonymous reporting and of protections from liability to encourage information sharing among members.

"HHS should provide the necessary support for the development and dissemination by professional organizations of additional standards and guidance for applying genetic tests in clinical practice. CMS should work with professional organizations to develop interpretive guidelines to enhance inspector training and laboratories.

"Recommendation 3. There are considerable information gaps about the number and identity of laboratories performing genetic tests and the specific genetic tests being performed. To gain a better understanding of the genetic tests being offered as laboratory-developed tests and to enhance the transparency in this field, SACGHS reviewed proposals for a voluntary or mandatory test registry and considered the benefits and burdens of each type of system.

"The community cited that a mandatory, publicly available, Web-based registry that is well staffed to maintain an accurate and current database would offer the best approach to address the information gaps. Since genetic tests are not unique from other laboratory tests for oversight purposes, the registry should include all LDTs. The Committee also discussed whether such a database should reside at CDC, CMS, or FDA.

"Based on exploratory work, SACGHS concludes that a mandatory registry should be established. The Committee recognizes that there are unresolved issues, including practical and

legal questions, that require further analysis before a final decision can be made about how and where to implement the registry.

"HHS recommends the following course of action:

"A) HHS should appoint and fund a lead agency to develop and maintain the mandatory registry for LDTs. The lead agency should work collaboratively with its sister agencies to create a comprehensive registry and minimize duplicative collection of registry information. The lead agency should have qualified personnel who are experienced in developing and updating large databases in a timely and accurate manner.

"The lead agency, in collaboration with its sister agencies, should convene a stakeholders meeting by September 2008 to determine the data elements associated with analytical validity, clinical validity, clinical utility, and accessibility of data that should be included in the registry.

"The lead agency should cast a wide net for a broad stakeholder representation, including representatives from the private sector who can represent a role for public-private partnership in developing the registry.

"The lead agency, through the stakeholders effort, should assess the level of effort as well as the burden on the laboratory and the impact on other key stakeholders such as patients, physicians, and payers, necessary to obtain each data element, including linking to reliable sources of existing information."

DR. KHOURY: Just, a registry for LDTs or all genetic tests?

DR. TEUTSCH: It should be genetic tests. You will notice copy edits and things like that which we are going to need to try and take care of. I don't think we need to do that as we go through this. So we will talk about that at the end.

DR. GUTMAN: I was going to keep my mouth shut, but you do have an opaque statement about exploring legal authorities. I'm certainly not a lawyer and I hardly can represent the FDA, so I certainly couldn't represent CDC or CMS or HHS in general. But there actually is at least a possibility that in order to do this there would need to be some statutory change.

DR. FERREIRA-GONZALEZ: Yes, we understand that.

DR. GUTMAN: It makes it awkward to have an October '08 meeting if you --

DR. FERREIRA-GONZALEZ: No, the October is for the stakeholders to gather the information on the elements.

"C. While awaiting completion of the other processes, HHS should use short-term voluntary approaches such as incentivizing laboratories to register with Gene Test and encouraging laboratories to make their test menus and clinical validity data for these tests publicly available on laboratory websites.

"Recommendation 4. There has been much debate in the past decade regarding FDA's role in regulating laboratory-developed tests. SACGHS supports FDA regulation of LDT and the" --

MS. ASPINALL: Excuse me. I'm not sure that is correct.

DR. FERREIRA-GONZALEZ: Yes, I was going to say.

[Pause.]

DR. FERREIRA-GONZALEZ: It was the clinical validity. There we go. That is the one.

"The Committee is concerned by the gap in oversight related to clinical validity. The Committee believes that it is imperative for this gap to be closed as expeditiously as possible. To this end, the Committee makes the following recommendations:

"All laboratory tests should be addressed by the FDA in a manner that takes advantage of its current experience in evaluating laboratory tests. This step by HHS will require commitment of significant resources in order to avoid potential harms (patient and public health, staffing, technological innovation.)

"The Committee recommends that HHS convene a multi-stakeholder public and private sector group to determine the criteria for risk stratification and a process for systematically applying these criteria. This group should also consider new and existing regulatory models and data sources, such as New York State. The multi-stakeholder group should also explicitly address and eliminate duplicative oversight procedures.

"To expedite and facilitate the review process, the Committee recommends establishing a registry as noted in Recommendation 3.

"Recommendation 5. SACGHS fact-finding also identified gaps in the enforcement of existing regulations. To prevent laboratories from performing genetic tests without appropriate CLIA certification, the following steps should be taken.

"The CLIA program has an array of enforcement actions available, but those actions cannot be imposed on an uncertified laboratory. Instead, CMS must report the laboratory to the HHS inspector general for action. Laboratories without CLIA certificates cannot be reimbursed by Medicare or Medicaid, but this restriction has no consequence for laboratories that perform direct-to-consumer testing.

"CMS should exercise its regulatory authority to take enforcement actions against laboratories that perform genetic tests for clinical purposes without proper CLIA certification. CMS should step up its efforts to make publicly available a list of laboratories that have been cited by the CLIA for condition level deficiencies."

DR. KHOURY: Andrea?

MS. ASPINALL: I have a question. Is this another one where we take out "genetic"? Because we just talked about all tests. Even if we don't define genetic, if a lab is doing a test.

DR. FERREIRA-GONZALEZ: Yes.

DR. KHOURY: Andrea, do we need the section in green? Can we just take it out? I always find it confusing. We have to acknowledge it, but it interrupts the flow. Maybe we should move it elsewhere.

DR. TEUTSCH: We can take care of that in editing.

DR. FERREIRA-GONZALEZ: That is in editing.

"Appropriate federal agencies, including CDC, CMS, FDA, and FTC should strengthen monitoring and enforcement efforts against laboratories and companies that make false and misleading claims about genetic tests."

DR. TUCKSON: Including the DTC.

DR. FERREIRA-GONZALEZ: Including the DTC.

DR. TUCKSON: You don't have to type it now.

DR. FERREIRA-GONZALEZ: "Recommendation 6. SACGHS is concerned about certain types of health-related genetic tests that are marketed directly to consumers and appear to fall outside the scope of CLIA. Some nutrigenomic tests, for example a test for caffeine metabolism, and tests to determine the gender of a fetus are examples of health-related genetic tests that are skirting the boundaries of CLIA authority. There is insufficient oversight of laboratories offering such tests and their potential impact on the public health is an increasing concern.

"SACGHS recommends that CLIA regulations or, if necessary, CLIA statutory authority, along with FDA risk-based regulatory authority and regulatory processes, should be expanded to encompass the full range of health-related genetic tests, including those offered directly to consumers. Relevant agencies such as CMS, CDC, FDA, and FTC, should collaborate in an effort to develop an appropriate definition of health-related genetic tests that FDA and CMS could use as the basis for expanding their scope."

DR. LICINO: Question. When you say they are health-related genetic tests, could people use that as a loophole and say this is just information about your biology, who you are?

DR. FERREIRA-GONZALEZ: This is what we are trying to close. That is exactly the loophole we are trying to close.

DR. LICINO: But then, should you take the "health-related" there and just say "genetic tests"? Because all of them have some health relevance.

DR. FERREIRA-GONZALEZ: No.

DR. LICINO: It could be like a genealogy. Let's say if you do the $1,000 genome thing comes there and becomes available, then they say, okay, I'm doing it to see where I come from, but the information is sequenced and can be used any way. Could people use that as a loophole and say I'm not doing it for health-related reasons, I'm doing it just to get a genetic history?

DR. WILLIAMS: I think that is why the point of the last thing, which was to develop an appropriate definition. Again, I don't think we can necessarily do that, but we can't leave it to the company to make the definition. That definition has to be defined by the agencies that we want to have regulatory authority.

DR. FERREIRA-GONZALEZ: The exact example that you described could be brought in, too. Martin.

MR. DANNENFELSER: But, a gender-related test may not be health-related. So I don't know that if that language applies. I think the idea of taking "health-related" out makes sense.

MR. MILLER: So, can you just take out "health-related" and just say certain types of genetic tests that are marketed?

MS. ASPINALL: Isn't it just tests? Again, this definition is moving, technology moves. I think some of those tests are being used in health-directed ways. I think the second sentence makes sense.

DR. FERREIRA-GONZALEZ: "Genetic tests."

MS. ASPINALL: Just certain types of tests.

DR. FERREIRA-GONZALEZ: Certain types of tests that are marketed directly to consumers.

MS. ASPINALL: It is consistent with what we said in the rest.

DR. FERREIRA-GONZALEZ: But then, "should collaborate in developing an appropriate definition of health-related." We leave that there.

DR. FROHBOESE: It is repeated again at the bottom, as well. The last line. "Health-related genetic tests."

DR. FERREIRA-GONZALEZ: Are we okay with this? We can work through that. That is a detail.

Next one. Chapter 5. Recommendation 1. "Information on clinical utility is critical for managing patients, developing professional guidelines, and making information on clinical utility of genetics tests. There is inadequate data on which to base utility assessments and only a few studies have been done of the clinical utility of specific genetic tests.

"More fundamentally, insufficient analysis has been done on the standards of evidence upon which the clinical utility of genetic tests should be evaluated, and evidence-based methods applicable to genetic testing have been developed.

"Further, policy analysis is also needed to define the process by which clinical utility assessments will be applied. To fill these needs, SACGHS recommends the following:

"HHS should create and fund a sustainable public-private entity of stakeholders to assess the clinical utility of genetic tests. An example is building on CDC's Evaluation of Genomic Applications in Practice and Prevention, EGAPP, Initiative.

"This entity would identify major evidentiary needs; establish evidentiary standards and level of certainty required for different situations such as coverage, reimbursement, quality improvement, and clinical management; establish priorities for research and development; augment existing methods for assessing clinical utility as well as analytical and clinical validity, such as those used by EGAPP and the U.S. Preventive Services Taskforce, with relevant modeling tools; identify sources of data and mechanisms for making them usable for research, including the use of data from the electronic medical records; recommend additional studies to assess clinical effectiveness; achieve consensus on minimal evidence criteria to facilitate the conduct of focused,

quick turnaround time of systematic review; increase the number of systematic evidence reviews and make recommendations based on their results; facilitate the development and dissemination of evidence-based clinical practice guidelines and clinical decision support tools for genetic/genomic tests; establish priorities for implementation in routine clinical practice; and publish the results of these assessments or make them available to the public via designated HHS or other publicly supported, like Gene Test, websites.

"To fill gaps in our knowledge of analytical validity, clinical validity and clinical utility, utilization, economic value, and population health impact of genetic tests, a federal or public-private initiative should develop and fund a research agenda to fill those gaps, including the initial development and thorough evaluation of genetic tests and the development of evidence-based clinical practice guidelines for the use of those tests, and disseminate these findings to the public via designated HHS or other publicly supported websites, such as Gene Test.

"Recommendation 2. Healthcare payers are increasingly requiring evidence of clinical utility before they will pay for genetic tests. Therefore, coverage and reimbursement decisions play a critical role in stimulating innovation and facilitating access to genetic testing. In February 2006, SACGHS issued a report that made recommendations for developing evidence of clinical utility and addressing other barriers to the coverage and reimbursement of genetic tests and services in the public and private sectors. SACGHS offers the following recommendation concerning the development of clinical utility evidence.

"As the issues identified in the Coverage and Reimbursement of Genetic Tests and Services Report ar still current, SACGHS urges HHS to act on the report's recommendations.

"In addition, public and private healthcare payers, in collaboration with relevant groups such as test developers and clinical laboratorians, should develop mechanisms such as development of phased reimbursement to facilitate the collection of clinical utility evidence for high-priority tests and applications. Implementation of innovative approaches should be accompanied by careful evaluation to assess whether they enhance or hinder innovation, understanding effectiveness, and appropriate utilization.

"Recommendation 3. The value of genetic tests to patients is realized only when they are used appropriately. In addition, quality improvement processes are needed to assure that genetic tests are delivered consistently to appropriate patients. Furthermore, an ongoing process is needed to identify opportunities for improving the use of genetic testing, including the collection of post-market outcome data. SACGHS therefore makes the following recommendations.

"HHS should conduct public health surveillance to assess surrogate and health outcomes practice measures, including the proper utilization and the public health impact on genetic testing. Information should be linked to quality improvement practices that affect patient outcomes and the provision of health services.

"Data on specific genetic testing results will be required to permit understanding of the significance of genetic variance and new detection methods to improve the utility of testing.

"Recommendation 4. The clinical utility and value of genetic testing is inextricably linked to methods to improve care processes and decision support. Interoperable electronic health records will play a central role in the translation of guidelines into care practices through their decision support and educational functions. They will serve as a critical resource for assessing clinical utility and quality of care. SACGHS therefore makes the following recommendations.

"HHS should ensure the coordination and implementation of efforts, including the deliberation of SACGHS and AHIC, or its successor, and other workgroups addressing personalized health care, population health and clinical care connections, and confidentiality, privacy, and security to advance the appropriate use of patient-level data for research and for enhancing the quality of decision-making.

"Chapter 6, Recommendation 1. There are documented deficiencies in genetic knowledge in all relevant stakeholder groups. In addition to the creation of the SACGHS Education Taskforce, SACGHS recommends the following strategies to address these deficiencies.

"HHS should work with all relevant governmental agencies and interested private parties to identify and address deficiencies in knowledge about appropriate genetic and genomic test applications in practice and education of key groups, such as healthcare practitioners, public health workers, public and private payers, and consumers. This educational effort should take into account the differences in language, culture, ethnicity, and perspectives on disability, as well as issues of medical literacy, access to electronic information sources such as the Internet, and deficiencies in public infrastructure such as libraries that can affect the use and understanding of genetic information.

"Based upon increased research regarding analytical validity, clinical validity, and clinical utility, sufficient resources should be provided for the translation of this knowledge into evidence-based clinical practice guidelines that enhance the quality of clinical care and public health outcomes. See also Recommendation 3, Chapter 5.

"Although FDA has asserted its authority over clinical decision support systems, the extent to which the agency intends to regulate such systems is not clear. Given that clinical decision support systems will be necessary to communicate information appropriately in the pre- and post-analytic period and because these systems contain elements that involve the practice of medicine, clarification of the nature and scope of FDA oversight of such support systems is critical.

"SACGHS recommends that FDA should engage with other relevant federal agencies, advisory committees to the Secretary of HHS such as AHIC and the Newborn Genetic Testing, and stakeholders to gather perspectives on the appropriate regulatory framework for clinical decision support systems in light of the change in healthcare delivery and healthcare data collection systems.

"As part of this process, FDA should prepare a guidance document articulating the basis of its authority to regulate clinical decision support systems, as well as rationale and approach to such regulation explaining in particular which features of the system constitute a device.

"The need for genetic expertise to support best genetic testing practices has been identified as an essential element for the provision and interpretation of appropriate genetic tests. Access to genetic expertise could be addressed in part by solving problems in the reimbursement of genetic tests and services. SACGHS recommends that HHS act on the recommendations of the 2006 SACGHS Coverage and Reimbursement of Genetic Tests and Services Report.

"There are extensive gaps in knowledge about genetic tests and their impact on patient care. Prioritizing activities under the authority of HHS would help to close these gaps and enhance the quality of patient care. SACGHS recommends that HHS allocate resources to AHRQ, CDC, HRSA, and NIH to design and support programmatic and research efforts in order to encourage

development and assist in the evaluation and dissemination of tools, particularly computerized tools, for clinical decision support in the ordering, interpretation, and application of genetic tests; and address current inadequacies in clinical information needed for test interpretation.

"These efforts will require engaging providers and payers as well as providing incentives and protections in order to ensure participation in design and dissemination of tools, implementation of clinical decision support, and contribution of necessary data."

DR. FOMOUS: The next one I think you want to decide if you want in or out.

DR. TEUTSCH: The next one we deleted, right?

DR. WILLIAMS: My sense was it should be deleted, but in the presentation of all the recommendations we wanted to assure that the Committee agreed with what I think, which is we have covered this already back in Chapter 4.

DR. FERREIRA-GONZALEZ: What about the privacy issues?

DR. WILLIAMS: We talked a lot about the need for this. We now have a recommendation in Chapter 4 that deals with the marketing of DTC tests, but it is general, as part of oversight of all of those marketing programs, including them, and doesn't specifically speak to this issue.

I think the preamble, which is the first paragraph, which is setting the stage, does present some unique aspects that probably need to be folded in, but the recommendation per se is covered. Basically, I think we need to work the text from the preamble into the relevant [section.]

DR. TEUTSCH: If people are all right with that, we will do that as part of the editing process and drop this recommendation because it is dealt with elsewhere.

Andrea, thank you very much. That is a real tour de force just to read.

MS. ASPINALL: We have one comment.

DR. TEUTSCH: We have one more? Oh, we have the overarching one. Did you read it or not?

DR. FERREIRA-GONZALEZ: Hold on, hold on.

DR. FROHBOESE: But, does the preamble adequately cover the issue of privacy?

DR. WILLIAMS: It covers it enough from the perspective of the recommendations. The text of the report has additional information on that. The setup for the recommendations is essentially just to give a little bit of a taste of what is actually in the report that is discussed in more detail.

MS. ASPINALL: I'm sorry. Can I go back to Chapter 6, Recommendation 3? I thought we were waiting until the end of the chapters. The issue that we have talked about several times and I think is critical -- so tell me if I'm missing something -- is the need for appropriate reimbursement for these tests.

DR. TEUTSCH: That is in this report.

MS. ASPINALL: It is in this report, which I understand, and I think it is great that we refer to it. I just read the preamble here as the need for genetic expertise focused on genetic counseling and other services but not the tests themselves.

DR. TEUTSCH: That is correct. This is the chapter on decision support and education.

MS. ASPINALL: That is where I was waiting until the end, but maybe it is appropriate here. I think it was the sense of the Committee to say it more globally, that in light of the importance of the tests and, quite frankly, additional requirements that we are going to have as a result of it, that we need to ensure that reimbursement is reviewed. That is not the FDA, and it is perfect for HHS, which has the CMS component.

DR. TEUTSCH: That is another recommendation, either No. 4 or No. 5.

DR. FERREIRA-GONZALEZ: Chapter 5. Do you want to go back to Chapter 5?

DR. TEUTSCH: We have referenced this report twice, once relating to tests, once relating to expertise.

MS. ASPINALL: Which one is it?

DR. FERREIRA-GONZALEZ: Chapter 5.

MS. ASPINALL: I didn't see it.

DR. FOMOUS: No. 5-2.

MS. ASPINALL: So that was why I wrote down 5-2. Can you just go back to the first piece of it? This, to me, was not about the overall reimbursement but rather to facilitate the collection of clinical utility evidence which was referenced in the other report, as opposed to more broadly.

DR. FERREIRA-GONZALEZ: Do you want to have a specific statement to also look at the reimbursement?

MS. ASPINALL: Yes.

DR. FERREIRA-GONZALEZ: Because it is covered in the report.

MS. ASPINALL: Right. I do. I am happy to have it anyplace, and maybe it is in the overarching place at the beginning, that this report is only complete in addition to the 2006 Coverage and Reimbursement Report. That would be fine. Nos. 5-2 and 6-3 are very specific and important, but it is fine if it is in the overarching. I was waiting until we saw these because they are very specific on individual issues.

DR. TEUTSCH: The whole report, Mara, on this topic was not a subject of great focus here except insofar as it dealt with oversight. We tried to allude to it at least on several occasions plus in the text. So we understand that that is an issue and that is, obviously, why we are continuing to address it.

MS. ASPINALL: I guess given that a lot of people will look at the recommendations, the two recommendations at 5-2 and 6-3 that mention it, mention it in reference to a specific aspect,

clinical utility or genetic counseling or genetic expertise. What I was saying at a higher level is this report needs to be connected with the Reimbursement and Coverage Report.

DR. FERREIRA-GONZALEZ: Where would you recommend that we put that? You want a third mention to this report.

DR. TEUTSCH: The other way to go is in the text. I think it is important that we do make these links because this relates to much of our work.

Could we ask you to take a look and make sure that it is clear in the text of the report that we are addressing the reimbursement issue, that it is taken up there, it is an important issue in making sure that there is access?

MS. ASPINALL: I think it is in the text. I'm saying it needs to rise to the overarching piece of the recommendations because I think it gets lost in 500 pages.

DR. TEUTSCH: Marc.

DR. WILLIAMS: I would make two suggestions. One is that we are obviously still engaged with the Secretary on the Coverage and Reimbursement Report, and many of the issues that are still outstanding there are being addressed. I personally don't think we need to keep beating this about the head and neck. I think it is relevant where it is in the recommendations. It is adequately addressed in the text.

I would suggest we just do a thumbs up/thumbs down in terms of how people feel about this. Is this something that is important to represent as an overarching? I would argue not. But if people feel strongly that way, then we can work on where to put it in.

DR. FERREIRA-GONZALEZ: Any language that we can add to any of these two recommendations?

DR. WILLIAMS: Not really, not really.

DR. TEUTSCH: Let me just get a sense of the group. How many people think we need either a recommendation or to modify something to add additional reference to the Reimbursement Report about the need for testing?

[Show of hands.]

DR. TEUTSCH: How many feel that we do not need to do that?

[Show of hands.]

DR. TEUTSCH: I take it the sense is we all recognize it is an important issue, and if we can strengthen it somewhere else we will have other ways to deal with that.

That is a real tour de force, Andrea. Let's see if we can get to a vote on the recommendations.

DR. FERREIRA-GONZALEZ: Are we doing the overarching?

DR. TEUTSCH: Do you want to go back to that?

DR. FERREIRA-GONZALEZ: Yes, the overarching.

DR. TEUTSCH: Read it.

DR. TUCKSON: Are you going to vote individually, Steve, or just can you try to vote en masse?

DR. FERREIRA-GONZALEZ: The overarching recommendation.

DR. TEUTSCH: We will. No, we just skipped over that and Andrea asked that we come back to it.

DR. FERREIRA-GONZALEZ: "The Committee found significant gaps in the U.S. system of oversight of genetic testing that could and do lead to harms. The Committee formulated a number of recommendations that, if implemented and sufficiently supported, could close these gaps. The Secretary of HHS is responsible for closing these gaps and fostering the public's health."

DR. FITZGERALD: I understand that there was some concern about the forcefulness of that final statement, but one can look at it in a different way that presents the same logic. Take that last sentence and imagine it in this form: "If these gaps are to be closed and the public's health fostered, then the Secretary of HHS must take responsibility for this process."

I think the fear is that this is coming across as some kind of determination by this Committee that the Secretary should take up some extra job or responsibility or something.

MS. ASPINALL: No.

DR. FITZGERALD: I think what we are trying to say is no, this is just part of the reality. If this is going to be done, the Secretary of HHS is the one to do it.

DR. TEUTSCH: Right. The Secretary, of course, has asked us to do it because, presumably, he believes that he has the responsibility for doing it.

DR. FERREIRA-GONZALEZ: Robinsue.

DR. FROHBOESE: I think, in keeping with that, aren't we missing something here? I'm wondering whether "responsibility" is the right concept or the fact that we need to tie the fact that the Committee formulated a number of recommendations. Don't we want to say that the Secretary can and should close these gaps by following these recommendations?

I think by trying to couch this in terms of responsibility we are missing that linkage. The Committee found gaps, formulated recommendations, and then the last thought is that the Secretary can and should close these gaps by following the recommendations.

DR. LICINO: Robin and Reed, both of you were not here. We had a discussion about this. The original language was "The Secretary of HHS must close these gaps," and then we discussed here how do we tell the Secretary what to do without sounding too presumptuous. That is the challenge, I think.

DR. TUCKSON: Because you are his advisory committee. He asked us to tell him how to do it. Whatever the discussion was, I don't feel a certain shyness here. The only word that I'm

concerned about is the word that we are saying these recommendations "could." I thought we felt pretty good these recommendations "will." We didn't come at this to say that there are big holes left.

So I think we should say that these recommendations would close these gaps, and I do like the way Kevin phrased it. I thought it was very nice.

DR. FITZGERALD: I don't think we can say "will close these gaps" because if they are not implemented they won't do it.

DR. FERREIRA-GONZALEZ: How about the language?

DR. TUCKSON: I think we want to just be real careful that you don't make it sound like you are undercutting your own recommendations by saying, hey, these are some nice ideas.

DR. FERREIRA-GONZALEZ: How about the last sentence?

DR. FITZGERALD: My understanding is the reason we put in that word was because Reed was pushing for accountability.

DR. FROHBOESE: But, can we get at it by saying "The Secretary "can and should close these gaps by"?

MS. ASPINALL: I like the wording. Because, otherwise, it is not connected to the other paragraph.

DR. FERREIRA-GONZALEZ: Can you repeat the wording?

DR. FROHBOESE: I'm recommending that we say "The Secretary of HHS can and should close these gaps by implementing the Committee's."

DR. EVANS: Or we can just put that last sentence as the second sentence and say, "The Secretary of HHS can and should close these gaps by implementing the recommendations that are contained."

DR. FERREIRA-GONZALEZ: "The Secretary can and should close these gaps to foster the public health."

DR. FITZGERALD: One thing I don't think we want to lose again is what Phyllis also pointed out. It is not just a matter of closing gaps. That doesn't solve all the issues. There are more. So we don't want just to worry about closing gaps. We want to actually move beyond that once that happens.

MS. ASPINALL: So, Kevin, with that, do we want to start with "The Committee found significant gaps and necessary improvements" or "potential improvements in the U.S. system"? You are right -- well, not just you, Kevin, but everybody -- it is not just about gaps that we are filling. It is about improvements.

DR. FERREIRA-GONZALEZ: Joe?

DR. TELFAIR: I'm very clear that you need to plainly state what it is that you want to do. To me, changing that, with all due respect, is not plain and is not straightforward. It is just saying this is your responsibility, this is what you do, and that is it. That is just laying it on the table. Also, it reinforces the fact that there are things that are beyond the main outcome, which is gaps. There are things beyond that that need to be done, and it is perpetual. It doesn't just stop at that point.

DR. FOMOUS: I think you need some sort of language in there that sounds like a recommendation because I think the prior sentence that we had in there was like this is all nifty and dandy, you are responsible. Where is the recommendation for action?

DR. TUCKSON: Maybe one way to do it is instead of saying "The Secretary of HHS can and should," "We recommend that the Secretary of HHS close these gaps." "We recommend that he close the gaps."

DR. FERREIRA-GONZALEZ: Yes, that's it. That's it.

MS. ASPINALL: What about gaps and improvements?

DR. FERREIRA-GONZALEZ: Hold on. Let's finish with this and then we will go back.

DR. TELFAIR: But there is an implied endpoint there if you just say "close the gaps." We already know that this is a perpetuating situation. Even though this Committee at this point in time has identified these, there are anticipatorily going to be other means that are going to come up. It was made pretty clear through both the public comment and in things we have heard that this is only basically the tip of the iceberg right now and there is more to come.

DR. TUCKSON: So maybe one slight way you could do it [is], "The Committee recommends that the Secretary of HHS close these gaps by implementing the recommendations and continuing to advance," "continuing to monitor and respond appropriately"? No, that's crappy. Never mind.

MS. ASPINALL: How about just "implement the recommendations"? We are giving recommendations. We are saying, listen, do it.

DR. EVANS: If you want to say something about the public's health, just say it at the end of the middle sentence. "Would close these gaps and foster the public's health. Period. The Committee recommends the Secretary of HHS close these gaps by implementing the recommendations."

PARTICIPANTS: No.

DR. EVANS: "Implement the recommendations." Period.

PARTICIPANTS: Period.

DR. FERREIRA-GONZALEZ: Mara, you had a comment?

MS. ASPINALL: I had a different comment from Sylvia's comment, I think. Somebody said it. The issue about "The Committee found significant gaps and potential improvements possible in the U.S. system." "Opportunities for improvement in the U.S. system." It is not as if it is all about gaps, it is also about improvements.

DR. TEUTSCH: Can we get a sense if folks are all right with this recommendation as it is?

MS. ASPINALL: Can we say "recommendations," period.

DR. FERREIRA-GONZALEZ: So, how about in the second sentence, "would close the gaps."

PARTICIPANTS: No.

MS. CARR: "Enhance the public health" after gaps? Second sentence.

PARTICIPANT: "Would close these gaps and enhance the public's health."

PARTICIPANTS: Yes.

DR. TEUTSCH: You can put that up there.

MS. ASPINALL: But I'm still going to come back with, are we really okay with saying the report is only about gaps?

DR. WILLIAMS: We are talking about enhancing the public's health. I think we addressed it.

MS. ASPINALL: How about, we are enhancing it through just --

DR. FERREIRA-GONZALEZ: "Enhance the system." We need to enhance the system.

MS. ASPINALL: We are just closing gaps, is what this sounds like. I think we are doing more than that.

MS. CARR: Enhance the oversight system.

DR. WILLIAMS: So you could say, "The Committee recommends that the Secretary of HHS enhance the oversight system to close these gaps."

DR. TEUTSCH: It should say "implement the recommendations." "Implement the recommendations and assume responsibility for improving the oversight of genetic tests." That is the ongoing part.

MS. ASPINALL: I think it just easier in the first sentence to say "close gaps and opportunities for improvement."

DR. LICINO: "To enhance" or "and enhance"? Would "close these gaps in order to enhance the public's health," right?

DR. FERREIRA-GONZALEZ: No, no. Paul. We need to finalize this.

DR. TEUTSCH: We need to get to closure. Paul?

DR. LICINO: You need "public health" twice. I like public health and all its advocates, but you need it twice.

DR. TEUTSCH: Paul.

MR. MILLER: The train may have left the station by now, but the title of this is "Overarching Recommendation," and the overarching recommendation is to implement the recommendations.

[Laughter.]

MR. MILLER: That just seems to be a little circular to me. What I thought originally, as I was thinking about an overarching recommendation, is in a sense going back to what, I forget whether it was Steve or Reed, said it. The loyal staffer comes in and says, "SACGHS has come up with this report." The Secretary says, "Well, that's great. What's the bottom line?" The bottom line is, in a sense, what is the real push here.

I like that first thing saying, hey, there are significant gaps and there is a way to solve these gaps. Again, the train may have left the station, but ultimately, what is the core thing, if you can capture it in a thought or a thing, in terms of the overarching point? Because of changes in technology and the greater use of genetic tests, more resources and more oversight and stakeholder input are needed to come up with something.

DR. EVANS: Closing the gaps, isn't that the overarching recommendation? Didn't we decide that, look, what has to be done? There are gaps. They have to be closed. Isn't that our overarching recommendation?

MR. MILLER: I'm just saying that this is an opportunity to say something more concrete, and I wonder if we are taking that opportunity. That is my point.

DR. TEUTSCH: I know we need to bring this to closure. I wonder if you could, with some forbearance, allow the steering committee to work on this and get it finalized.

DR. FERREIRA-GONZALEZ: We keep going around and around.

DR. TEUTSCH: I don't think we can wordsmith this by committee at this moment.

DR. FOMOUS: Instead of calling this an overarching recommendation, could we call it an overarching scene? Then there wouldn't be this sense that you have to be recommending an action to capture all of the recommendations.

MR. MILLER: I think you do need it. I think the report is stronger by having an overarching recommendation that follows from, we found significant gaps and here is something concrete that you need to do.

DR. TUCKSON: I think the suggestion from the chairman is that we have a sense of what is needed. The committee is going to have to work this one a little bit. It is not a technical recommendation. It is a matter of style here. I think Paul's comments are well taken here. I think the Committee understands it.

I would pile in that as you think about it offline that I realize that we have lost the key, which I think is the overarching issue, accountability. Alphabet soup. Americans are concerned. There is somebody in charge. Deal with it.

What I would suggest from a process point of view is that we have, I think, gotten the recommendations done. The Committee can grapple with this and send it back to us for wordsmithing or approval on the final.

I think we need to take a vote and get this done. It is almost one o'clock. That is our drop-dead date. We have four minutes before the end of the hour, and we have a critical quorum issue to deal with. So unless there is a whole big issue that somebody else has, we should vote.

DR. TEUTSCH: We are going to vote on the recommendations in each of the chapters. We are going to have the steering committee work on this overarching one. What I would like to do is to proceed to a vote as we have talked about. So, other than editorial changes and some wordsmithing on this, that is what we will be voting on.

Before we actually take our vote, we need to make sure that we have a record that two of our members, Paul Billings and Paul Miller, still have some pending paperwork, so that they are not going to be able to vote because of a delay in that process. Apologies to both of you.

For the remainder, we do need to take a vote. Do I have a motion in favor of accepting these recommendations?

DR. TUCKSON: I move.

[So moved.]

DR. WILLIAMS: Second.

[Motion seconded.]

DR. TEUTSCH: One from Reed, and a second from Marc. All in favor?

[There was a chorus of "ayes."]

DR. FOMOUS: Show of hands, please? And keep them up.

[Show of hands.]

DR. TEUTSCH: All opposed?

[No response.]

DR. TEUTSCH: Any abstentions?

[Motion carried.]

DR. LICINO: Can I make a motion?

[Laughter.]

DR. TEUTSCH: Go ahead, Julio.

DR. LICINO: I would like to make a motion for us to approve this as is without further editing.

DR. FERREIRA-GONZALEZ: No, the steering committee has an idea.

PARTICIPANT: It may wind up like that.

DR. TEUTSCH: It may. I get a sense that there is a significant amount to do there.

DR. TUCKSON: Mr. Chair, can I make a motion? I would like to make a motion that we give a resounding --

DR. TEUTSCH: This is what I was --

DR. TUCKSON: Oh, if that is what you were going to do, Mr. Chairman, it has more power coming from you.

DR. TEUTSCH: No. I think this has been an extraordinary effort on the part of Andrea and all of the steering committee, the taskforce members, the staff.

DR. FERREIRA-GONZALEZ: The staff.

DR. TEUTSCH: An enormous amount, and all of you.

DR. TUCKSON: And the public.

DR. TEUTSCH: And the public.

DR. TUCKSON: And the public, and the public, and the public.

MS. CARR: Implicit in what you just voted on is that you have also approved the spirit of the report going forward. Everybody understands that. You need to get edits by February 20th to Cathy.

PARTICIPANT: Yes. Can you just go through this process of edits by February 20th and then the completion by the 29th?

DR. TEUTSCH: The recommendations by the 29th will be finalized and sent to the Secretary. We will copy-edit the document and then the final version will go in April.

MS. CARR: By April. But we will copy-edit the recommendations and you will see what the steering group came up with on the overarching. You will see all of that again. But, no more edits to the recommendations.

MS. ASPINALL: Except as we have talked about them going through it because there were still some clarifications in putting them together.

MS. CARR: We won't be receiving more edits or seeking more edits from you guys on the recommendations.

DR. FERREIRA-GONZALEZ: We are just going to be cleaning up the language to assure that we have the "genetics" replaced and so forth.

DR. TUCKSON: I didn't hear the applause.

[Applause.]

DR. TEUTSCH: Thanks, everyone. That has been an enormous amount of work. We were going to take a 45-minute break for lunch. That will put us back here -- hopefully they are still serving -- at quarter to two.

[Lunch recess taken at 1:00 p.m.]

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