DIRECT-TO-CONSUMER GENETIC TESTING

Presentation of Draft Report on Direct-to-Consumer Genetic Testing

Sylvia Au, M.S., C.G.C.

[PowerPoint presentation.]

MS. AU: Thank you, Steve. First, I'm going to present what the taskforce has been doing in the three months since the last time we had our meeting. Then we are going to have some time for discussion of the draft paper.

For those of you who have been on the committee, you know that this is super speedy. We have never done anything this quickly, except for a letter. Sometimes the letters take longer than this.

I want to start by going through some of the background and intent of the paper, some of what we are saying in the paper, and the recommendations.

During the last meeting, we had established a short-term taskforce to look at direct-to-consumer genetic testing. The objective of the paper was to outline the benefits and concerns related to direct-to-consumer testing, highlight our prior SACGHS recommendations that might address those concerns -- we thought that might be a

good way of bringing back some of the concerns and recommendations that we had for other things to the new secretary -- and also identify issues that are not adequately addressed by our recommendations that we have made and that the committee might want to consider for future work.

Of course, as with all activities, we have a wonderful, educated, informed taskforce. A lot of these people owe me big favors for 4:30 a.m. conference calls. I was just telling Cathy, I should have scheduled a 4:30 a.m. conference call for you on the east coast just so I could do some payback.

Of course, I want to thank Cathy because she has done the lion's share of the work. She has been wonderful. For those of you who are new to the committee, we have the most wonderful staff of any committee ever. We want to keep that secret so no one steals them.

The goal of this session is that we are going to come to some consensus, hopefully some happy medium, about issues related to direct-to-consumer genetic testing, the prior recommendations that we want to bring forward to the Secretary that relate to this area, and any remaining

concerns that may require additional action by this committee.

Of course, we always try to limit the scope of our paper because we don't want to address everything under the sun. This direct-to-consumer genetic testing the taskforce decided would be limited to risk assessments, diagnosis of disease or health conditions, information about drug response, or other phenotypic traits. We excluded forensic analysis, ancestry testing, and paternity testing as much as we could. We also kept the definition of "genetic testing" from the Oversight paper, to be consistent. Because the recommendations from the Oversight paper address that definition, we didn't want to change it.

The intent of this paper recognizes that, of course, as usual, not all the concerns of direct-to-consumer genetic testing relate solely to direct-to-consumer genetic testing. They have great overlap, just like all our other papers do.

We also do identify issues that may be unique to direct-to-consumer genetic testing if a consumer's personal health provider is not involved in the testing. Sometimes government regulations that pertain to genetic testing may

not apply to direct-to-consumer genetic testing because of the way that the testing is done.

We will start with the benefits of direct-to-consumer genetic testing. The taskforce identified many benefits because, obviously, we know that there must be some reason that people would want to have direct-to-consumer genetic testing. We feel that it offers increased availability and access to genetic testing. It supports consumer empowerment and autonomy.

It promotes health literacy. That was one of the things that we discussed in detail because it would hopefully drive the consumer to learn a little bit more about genetic testing. It might drive their health care provider to learn a little bit more about genetic testing if there was direct-to-consumer genetic testing done.

It supports adoption of health-promoting behaviors, hopefully. If someone got a result that said that they were at higher risk for XYZ disease, they might change their health behavior to become healthier.

It provides an alternate route to medical research. There are research aspects to some of these companies, and that might be a route to research, as the

Parkinson's Disease Foundation told us about yesterday, that consumers might want to take.

It offers confidential access to genetic testing to those that might be concerned that there might be adverse action such as discrimination against them if the results were known.

So, our concerns about direct-to-consumer testing. The unprecedented speed at which the genetic technologies are involving and being translated into commercial products and then sold directly to consumers has raised definite concerns in the past for us. As in our Oversight paper, we do have concerns about test quality and analytical validity. We also have some consensus about a lack of standardized terminology for genetic variants, standards to select and validate variants used in assessing disease risk, and standard criteria in assessing aggregate risk. That we had discussed during our last meeting.

We have, of course, as we did in the Oversight paper, limited evidence of clinical validity and/or clinical utility of certain tests, particularly those involving risk estimates for common disease.

We also are concerned with false and misleading

marketing claims and incomplete or unbalanced promotional materials, those materials that might only reflect the benefits of what you might get from the genetic testing and not any of the down sides of it.

The ability for consumers to evaluate the marketing claims and make informed decisions about genetic testing is a concern, as well as the ability of the consumers to understand the test results once they get back to them, and the health care providers being inadequately trained or having inadequate knowledge to be able to help interpret those results once their patients bring in the direct-to-consumer genetic test results to them.

We also have limited data on psychosocial impacts on direct-to-consumer testing. We have concerns about protection for the research use of specimens obtained during direct-to-consumer testing and the data derived from the specimens.

There might be unclear or inadequate privacy protections because of the way direct-to-consumer testing might be provided to a consumer. There are inequities to access, of course, because you have to pay for the test in order to get the test. There are insufficient safeguards

to prevent non-consensual or third party testing. There are gaps in regulatory oversight, as we saw in the Oversight report, for genetic testing in general.

When we back over our old recommendations that we had made over the many reports that we have done, we found that there were eight recommendations from prior SACGHS reports that address some of the concerns that were raised. Of course, we found that there were some concerns that had no recommendations yet. Those are the ones that we will bring up for future consideration.

We had one recommendation on analytical validity, one on clinical validity, and one on clinical utility. Consumer and provider education had three recommendations. Companies that skirt regulations, one recommendation, and false and misleading claims, one recommendation.

I am not going to read our recommendations again in detail because for some reason our committee likes to make very wordy, long recommendations. You should all have this memorized, and the new members better have it tattooed on their bodies somewhere.

For analytical validity, of course, same as the Oversight report, we know that there are gaps in how

analytical validity and clinical validity data are generated and evaluated for genetic tests. We did recommend to HHS that they should ensure funding, which is a lovely recommendation that we always do. Ensure funding for the development and characterization of reference materials, methods, and samples. Methods to increase the analytical and clinical validity data, basically.

Continuing, for analytical validity again, funding for development of a mechanism to establish and support a laboratory-oriented consortium to provide a forum for sharing of information. The HHS agencies should continue to work with the public and private sector to support, develop, and enhance public reference databases with this information in them.

Again for analytical validity, we have that HHS should provide the necessary support for professional organizations to develop and disseminate additional standards and guidelines for applying the genetic tests in clinical practice.

On to clinical validity. We have the recommendation, again from the Oversight report, that the committee is concerned with the gap in oversight related to

clinical validity and the FDA should address that all laboratories should take advantage of its current experience in evaluating laboratory tests. This would probably require a significant commitment of resources.

Continuing with clinical validity, we have the recommendation that HHS convene a multi-stakeholder public and private sector workgroup to look at the criteria for risk stratification, process for applying use criteria, et cetera. Also, to expedite and facilitate the review process, the committee recommends the establishment of the much-beloved mandatory test registry that was a little controversial. Mainly the mandatory part was controversial, not the test registry.

Then, for clinical utility, again we have that HHS should create and fund a sustainable public-private partnership to assess the clinical utility of genetic tests. Then it goes on with a long laundry list that covers two slides on what that public-private partnership should do. I will not read every single one of those points.

Again for clinical utility, to fill the gaps in knowledge of analytical validity, clinical validity,

clinical utility, utilization, economic value, and population health impact, the federal, public, and private initiatives should develop and fund a research agenda to fill those gaps and disseminate those findings to the public via designated or publicly supported websites.

Then we get on to the education recommendations. Just like we talked about yesterday, the HHS should work with all relevant government agencies to increase training and education for all the key groups involved in genetics and genetic testing. That should be culturally competent, in many languages, et cetera.

The other one is to ensure that providers have appropriate education and training and are able to integrate genetics education into all areas of practice.

Continuing with our education recommendations, the HHS Secretary should provide financial support to assess the impact of genetics education and training on health outcomes and incorporate genetics and genomics into relevant initiatives of HHS, including the National Health Information Infrastructure, which I think that we talked about yesterday.

Patients and consumers should have information to

be able to evaluate health plan benefits so that they can figure out reliable and trustworthy information. Have federal websites with accurate information available to them.

Then we have our lovely CLIA and FDA recommendations. We recommend that CLIA would look at the regulations and hopefully, within their statutory authority, expand their regulatory authority to encompass the full range of health-related tests. Also, the FDA should exercise its regulatory authority to its full extent.

We have the recommendation that addresses false and misleading claims. Appropriate federal agencies should strengthen their monitoring and enforcement against laboratories and companies that make false and misleading claims about laboratory tests, including direct-to-consumer tests. We must have been very forward-thinking at that point to make that recommendation because it fits right into our report now.

So, we get to the part where the taskforce identified the concerns that we could not find recommendations that we have in prior reports that would

address those concerns. Some of those concerns that we might want to consider for future action are the concerns about unclear or insufficient privacy protections, limited data on psychosocial impact of direct-to-consumer genetic testing, potential exacerbation of health disparities, and inadequate protection for research use of specimens and data derived from the specimens.

I think that mainly came about because there would be certain entities that might not be covered under an IRB because they are not federally funded. What if they just decided that they didn't want to follow any of the federal regulations for research.

The lack of standards for genetic variant terminology, selection and validation of variants used in assessing disease risk, and calculating aggregate risk from multiple variants, is another issue that the committee might want to take up.

Today what we would like to do is have you tell the taskforce, are the issues related to the use of direct-to-consumer genetic testing addressed in this paper adequately? Do our prior recommendations address these issues? Are there any of the remaining concerns, and maybe

some new ones that you might identify, that might require additional action from the committee?

Finally, our next steps are to decide whether this paper should move forward to the Secretary of Health and Human Services. If we do decide to move forward, we will have to decide what the timeline will be for the edits and when we will transmit the paper, and determine what additional action the committee might want to take on some of the concerns that have not been adequately addressed by prior papers or recommendations.

Now we will open it up to complete agreement from the committee and move on. Opening the floor now to anyone that has any questions or comments? Yes, Marc, of course.

Committee Discussion

DR. WILLIAMS: First of all, I think you did an excellent job. I think taking the recommendations that are relevant from previous statements that have been vetted is the way to go. I read through the statement. I really didn't have any concerns or issues. I think that even as it is, recognizing that there are some issues that may not have been adequately addressed, I think it is appropriate to move forward.

The only thing I would add to the laundry list of things that have not been adequately addressed by previous recommendations would be the issue of sample and data ownership. One of the other things that has come up with the direct-to-consumer testing is, if a company was sold to another company, what would be the rules around transfer of those specimens, ownership, that type of thing. That is another area where there don't appear to be explicit protections relating to the consumer and how that information could be used.

That would be the only thing I would add to that bulleted list of things that we might want to consider doing more.

MS. WALCOFF: This is a very big area. I just want to make sure I'm understanding the process. From this point we would go back and take a look at these and the prior recommendations and really scrub them to make them more relevant and updated? How does that work?

MS. AU: We didn't want to change any recommendations because most of the recommendations here fit within the general topic of what we are talking about for direct-to-consumer testing. The new recommendations

that might need to be made then would take longer.

What we really want to do is move this quickly because, if we are making new recommendations, it generally takes a very long time. Even though it is not directly aimed at direct-to-consumer genetic testing, the scope of the recommendations fits the concern of direct-to-consumer genetic testing. Then if the committee decides that we need to hone in more, then those would be new recommendations that then we would decide to move forward to make.

MS. WALCOFF: I have a couple of thoughts on that. First, I think there is a lot of confusion between direct-to-consumer advertising and direct-to-consumer genetic testing and physician-ordered testing. I don't feel like taking the recommendations that apply to all things that we have done in the past really addresses the issue as well as we perhaps could. I think that this is something that people are paying very close attention to and are looking for more specific advice with respect to direct-to-consumer genetic testing.

Also, just generally, I think if we are going to provide advice to the Secretary my recommendation is to

update some of these recommendations in a way that is more useful to the Secretary. Hopefully they will get more attention and actually be implemented. I think it is very difficult with something like "HHS should ensure funding for." They don't really know what to do with that.

I know it sounds great and it is important, but I think it is better if the Advisory Committee can really give advice that can actually be implemented. I know we would like to give rapid advice, but it doesn't help if we get it there and then it just sits on the shelf because it is impossible or incredibly difficult to implement.

I would propose that we would go back through these and really direct this issue to direct-to-consumer genetic testing and really walk through these again to see how we might reformulate them. Maybe that is a strong word for trying to redo these. They were recommendations that were made before on a broader aspect of testing, but give the Secretary some more directed recommendations that can be more valuable more immediately.

DR. TEUTSCH: Are you suggesting that we go back and reassess all of these in terms of genetic testing and actually do the kind of reviews that led up to those

recommendations? Is that what you are suggesting, or just that we rework the recommendations themselves?

MS. WALCOFF: I'm not trying to add so much more on. That is why I'm not sure exactly what the process is in terms of where we are at this point with this. My understanding is these are all from reports previously that are broadly across the genetic testing landscape?

MS. AU: They are from different reports, not only Oversight but we have the Coverage and Reimbursement report.

MS. WALCOFF: So these have already been made.

MS. AU: Yes.

DR. TEUTSCH: Correct.

MS. WALCOFF: That is my point. I don't know to what extent they have been implemented or not, but if we are going to be making new recommendations or recommendations generally on a more specific area of direct-to-consumer testing, I don't know that it is that valuable to go back and just plug in the older recommendations.

It might be more valuable to take a little bit more time to get a short list of things that would be

directly associated with where the concerns are and focus on direct-to-consumer advertising. Take from the concerns and the recommendations that were addressed before but make them a little more directed and specific.

DR. BILLINGS: I completely agree with Sheila. First of all, Sylvia and the staff have done a masterful job of pulling this together.

MS. WALCOFF: Yes, it is a lot. It is a challenging area.

DR. BILLINGS: Pulling this together at all. For instance, on this whole issue of education, we identify DTC as potentially improving education literacy but also being misleading. Then we say we should fund better genetics education. It seems a little unrefined as a recommendation and also difficult fundamentally to implement. I do think we can edit it down and make the linkages to DTC a little more explicit.

MS. AU: I think this is an interesting topic in the news since Amway is getting into it now, according to what you forwarded me yesterday. We have our local Amway rep that will be doing direct-to-consumer genetic testing.

I think this was thought of as a vehicle to bring

up recommendations that were general and that crossed a lot of areas to the new Secretary. Besides the summary of what we have done, this will be the first issue that is brought up to the Secretary.

I don't know what the taskforce or the committee thinks about going back and narrowing all the recommendations down because they aren't really specific to direct-to-consumer testing. If we recommend education, it crosses the board because we have a whole Education Taskforce that is doing that.

MS. WALCOFF: Right. I guess that is my question. Are we making recommendations in a report on direct-to-consumer genetic testing or just pointing out all the various recommendations we have made across the board generally to her.

MS. AU: I think what we are doing is we are describing the issue and then the recommendations. Here are our prior recommendations that are still in effect that would address the concerns of direct-to-consumer testing. That would fit.

DR. NUSSBAUM: Sylvia, first of all, I think you and the taskforce have done an extraordinarily

comprehensive job. That is the applause.

Like Sheila and others, I believe that this really needs more of a focus on the DTC issues. First, people understand DTC. We have seen it arise I think very significantly in the pharmaceutical industry when claims have not been always backed up by science. I think that should be the paramount focus.

If you do that, then under that theme we can bring some of the issues of consumer knowledge and education. You can bring in some of the themes of clinical validity and scientific themes. I think what this document does is covers too broad a landscape, so much that focus would be lost. If you do focus on DTC, the issues that came up yesterday on the integrity of how samples would be used and consent and all of those issues, are really very relevant.

I think there is another dimension that one could work along, and that is where Marc was going. These are very early-phase companies. They don't have a strong financial backing in many cases. What happens to samples and what happens to information when they don't succeed. I think those are some of the safeguards that need to be

built.

Like my colleagues here, I think we can absolutely focus on DTC, the safeguards, the clinical validity and the claims that are made, and then build around it, but right now we just paint such a landscape picture that I think it is less actionable than it could be.

DR. ROYAL: I would just say details, details, details. Great job, Sylvia. I think your group has really brought the issues together.

I do agree that in moving forward those points that you made that are future might be ideas for future recommendations. I think you could focus on some of those. The impact of health disparities, the psychosocial impact of the information, a lot of those have not been addressed. Rather than leave them as potential future recommendations or topics that we may want to work on in the future, I think focusing on some of those might be where we could bring something new to the discussion.

MS. AU: I just want to remind the committee that last time we presented this as the outline for what we were going to do as a short-term taskforce. If we move to redo

recommendations, add new sections, it is going to expand the scope of what this project is going to be. If that is what the committee wants to do, then I think we have to make some decisions based on staff and other resources.

I just want to remind the committee that this was not the outline that was addressed last time.

[Laughter.]

DR. NUSSBAUM: On the other hand, you are also getting a new set of eyes on this. An extraordinary body of work has been achieved here, but how do you make it more meaningful. That is what I think we are all trying to drive to.

DR. WILLIAMS: I'm seeing a blended view here. One of the key points, in my view, is that the companies in many cases are trying to separate themselves out by saying, we are not doing genetic testing, we are doing education, or we are doing recreation, or we are doing something but we are self-defining this as not being genetic testing.

I think we can very rapidly say, you are doing genetic testing and in fact you are subject, or should be subject, to the same oversight that anybody else doing genetic testing is subject to. Therefore, the

recommendations from the Oversight report I think are extremely relevant to the direct-to-consumer things.

Now, I don't know that we necessarily have to fully recapitulate them, but I think it is important, given that we do have a new secretary, to say these things are very specific to them. That would be something that could be done in the short term.

In the medium term, I am resonating with some of the voices to say there really are some unique issues to direct-to-consumer, most of which have been outlined in your bullets, that probably do deserve some more study. The problem that I think we will encounter, as we did with Oversight, is just how much data is out there to actually be able to synthesize. I think in the long run it is going to come down to a lot of gut feeling about it.

Perhaps even a white paper that highlights the issues about what do we know, what do we not know, and what are the existing standards around research where maybe these are falling short, is worth some additional investment and time. It would certainly not, I wouldn't think, be worth the investment of doing a full report, but it is probably worth a little more effort.

That would be my recommendation, to go forward with the things that we know well that are relevant relating to the genetic testing aspects of it and the oversight of that. Then, make a more tailored document relating to some of the things that do appear to be more unique to direct-to-consumer testing.

DR. EVANS: I think maybe we can reconcile the old with the new by taking a page from a discussion we had yesterday. I will just put this out there.

I think that perhaps what we ought to do is draft, again, a very short document, a one- or two-page document, that says in a preamble something about how DTC is getting a lot of attention and we have some concerns. We are including as an appendix work that the committee has already done which addresses a lot of these issues, but here are bullets of, say, three things that we feel need to be on your radar screen. Maybe four.

We could, I suspect, pick a few of the things around the table, some of which have already come up, that rise to that level. I would put out there two things. To me, clearly the most important issue in the whole DTC arena is reconciling claims with reality. We address that in

here, but I think it could rise to the level of here is a bullet on that first page.

I would expand just a little bit from what Marc said. I don't think the issue is so much genetic testing as it is medical testing. If people want to get their earwax type from 23andMe, be my guest. When they are doing, as they are, the Ashkenazi founder mutations with high penetrance for breast and ovarian cancer and then claiming that this isn't medical testing, that is clearly in Congress. We should pick a very limited number of such things, put it in a front piece, and then I think we could as an appendix say, here is stuff we have done as a committee that addressed this.

MS. AU: So, you are suggesting the short letter, the previous work, and then taking the paper and expanding it to --

DR. EVANS: No, I'm saying a short front piece that says here are the three bullets that rise to the level and attached is also work the committee has done and now extracted from prior work that addresses this general topic.

MS. AU: That would be the recommendation part.

The front part of the paper is actually describing the whole area.

DR. EVANS: I actually would say have it all preceded by a one-page document with a very brief preamble that says here are some issues about DTC that rise to the level. Here is also a report that we have done that gives you background and extracts what we have done in the past. Does that make sense?

All I'm advocating is over-layering the whole thing with an executive summary that has a few bullets that we can decide around the table, probably in fairly short order, rise to the level of look at this. I suspect most people don't read after the first page if they see the executive summary.

MS. WALCOFF: I would be interested, too, to see what sort of specific recommendations those would be. If the biggest issue you see is the definition of testing --

DR. EVANS: And what the claims are.

MS. WALCOFF: -- is the next step all of these tests should be run through CLIA-certified labs? I think that is the next thing. I don't know what we would say about that because they should be defining them in some

way. Here is what HHS can do.

DR. EVANS: That particular example gets at two separate things. CLIA certification corresponds more to issues like analytical validity whereas reconciling claims with reality gets to more the oversight of the FTC, FDA, et cetera.

MS. WALCOFF: Right. My point is that we be more specific like that. We have all the agencies here that can give this input on what they can do, what they have been doing, what they could do, who they could partner with. Is CDC doing some of this under EGAPP. Is FDA doing some of this already. We could really assist them in getting attention for those efforts, but also, as we have talked about, defining them towards direct-to-consumer advertising.

We do have a new Secretary and new staff. There is a lot of publicity about these types of tests, where there is less publicity about when you are having a baby and you go in and have prenatal testing. I think it is important to have the report part to help define that for staff and others that want to go back in and delve, and to highlight the work that the committee has done before.

It is a vast amount of work. Sometimes, as I said, that gets lost in the transition of new people into new offices. I think that does help in preventing to reinvent the wheel and the work.

We have identified issues that you have identified that are important. Perhaps we can, in the shorter term, hone in on several of those and say we recommend, Madam Secretary, that you have some focus on this, you direct your agencies to focus on this sooner rather than later. These are other things that could apply generally.

MS. AU: I think Alberto, of course, wants to jump in.

DR. GUTIERREZ: I actually think that defining these as medical devices would be very helpful. That puts the onus then on the agencies to deal with them.

It also may be good to perhaps let the Secretary know that there are issues as to what laboratory-developed tests are or are not and what the different agencies are doing with them that need to be dealt with in one way or the other. It is public now that there is, at least within DFDA, a petition for us to deal with laboratory-developed

tests as regular tests, so that is something that the Secretary can look into and deal with as part of the issues that need to be dealt with.

MR. BOWEN: One particularly strong point of the report that I think would be good not to lose in terms of emphasis, and this leads back to education, is that it does a good job of delineating personal utility and clinical utility. We have found from our research that those two things are often confused by the public and policymakers. Clinical utility is not in the eye of the beholder. I just thought that was a strong point in terms of the education point.

DR. BILLINGS: I want to voice my support for something along the lines of what Jim said. I think Sheila and Jim were more or less arguing for the same thing.

I'm not aware that we have ever decided that direct-to-consumer testing was a medical device, so I have lots of concerns about that. I just want to be clear on that, at least as a member.

I wanted to just make two specific points. One of the things that distinguishes direct-to-consumer from other kinds of medical testing or genetic testing is the

role of the expert in ordering the test. That is not addressed in this document at all as far as I can tell. Maybe I missed it, but it is certainly a key distinguishing characteristic.

The direct-to-consumer folks say that this of course adds to access and empowerment and all those other things, but we might actually recommend or say something about that difference. It is an interesting issue for study, frankly, whether there is a benefit and the harms of not having the expert deeply inculcated in the actual making of the test menu.

The second point that I wanted to make was around the issue of privacy and so-called protections derived by direct-to-consumer access to testing. I think it would be quite valuable to have a box or some sort of opinion as to whether in fact there are any real protections derived by ordering a test through a direct-to-consumer pathway that are different.

As I remember, and I studied this a few years ago, if that information is subpoenaed, they have to produce it. Now, if it is anonymized in some way where it is impossible to get to the information, okay. Basically,

they are governed by the same laws as every other kind of testing. That was my impression, but I think we ought to say something definitive about it in the report.

DR. TEUTSCH: Let's make sure that we are all on the same page. These tests, to the extent that they have some clinical utility, are medical tests. Is there agreement about that?

DR. EVANS: A subset are, yes.

DR. TEUTSCH: The ones that have clinical utility.

DR. EVANS: I don't even think you need to say that because there are many of these tests that have clear medical implications but no demonstrated clinical utility.

DR. TEUTSCH: Right. Medical implications.

DR. GUTIERREZ: I would suggest that you actually say "that make medical claims."

DR. EVANS: Yes, yes.

DR. GUTIERREZ: That is what you want to say.

DR. FERREIRA-GONZALEZ: They can claim that they don't make clinical claims.

DR. EVANS: They can claim they don't make claims, but they are making claims.

DR. FERREIRA-GONZALEZ: I understand.

DR. TEUTSCH: When we talk about medical, that is about risk reduction. Health claims, basically.

DR. EVANS: There is no way you can reconcile the offering of high-penetrance LRRK2, BRCA, or mutational testing with the statement at the bottom of every page which says this isn't medical advice, it is not meant to diagnose, to treat, to recommend. They are just incompatible.

DR. TEUTSCH: Even risk prediction and other kinds of things that have behavioral implications for health, they would be included, correct?

DR. EVANS: Although, again, they also do testing that isn't medical.

DR. TEUTSCH: I understand. If you are doing ancestry, it is something different. I want to make sure that everybody in this room is on the same page with this, or at least that there is an overwhelming consensus, because that is actually a powerful statement that we have not made before. That then gets us into all of these other things. They need to have the same type of oversight, and then we can get into the kinds of things that relate to

unique characteristics of these things that I'm beginning to hear. Is that where people are?

MS. AU: How about the testing for vitamin use?

DR. TEUTSCH: You are making a health claim. It would be.

MS. WALCOFF: Do we have access to the NIH Counsel's Office or something like that? I think it is important, if we are starting to create new words or new definitions, what does that mean in terms of the existing statutory and regulatory framework. What are we trying to get at with that.

I don't know if we are trying to recommend that we parse these companies and say here is what we are going to say you should define as a health claim versus this. Are we going to be that specific? That is the only thing that is coming to my mind as an example. Should these tests be performed in a CLIA-certified lab? What are we trying to get at with creating a new terminology?

MS. AU: It is not creating new terminology. We are trying to limit that what we are addressing are the tests that make medical claims.

DR. TEUTSCH: Yes. I don't think we are trying

to create new terminology.

DR. EVANS: What I keep coming back to is, what we want to have rise to a very prominent position in our discussion, recommendations, knowledge in the Secretary's mind, is the medical claims being made and that there needs to be a reconciling between the claims made by the company and what they are actually doing. That is all. I don't think we are invoking new terminology.

DR. TEUTSCH: I think what we are saying is that the standards for the DTC, when you are making a health kind of claim or indicating some value in the health sphere, need to be at least as high as they are for when they are doing in the clinical arena. In fact, the reason things are doing in the clinical arena is you have a learned intermediary. That is gone. That is what Paul was getting at. That learned intermediary is gone. They are, in some sense, less capable of making a judgment about the appropriateness of the test.

DR. BILLINGS: Differently enabled, I would say.

DR. TEUTSCH: Differently enabled. We need to make sure that the information available to them is at least as good as what you would have in a clinical arena.

That is what I'm hearing here.

MS. AU: That doesn't mean that there aren't concerns when you do ancestry testing or match-making. They can still hold your genetic information and sell it or whatever. It is just that we are trying to draw a box around what we want to make recommendations about and not about the ancestry testing or that more recreational match-making and things like that. They do have concerns.

I think Paul had something to say.

DR. WISE: Thank you. I think what Jim is trying to have us do with the process of coming up with the one-page, three-bullet memo is to address the questions that people really have about DTC that are not directly addressed in this report. Crystallize the things that really are on people's minds. This issue is one of them. Is this a medical test or not.

My concern is that we could all sit around the room here and generally agree, but it is a fairly important decision and things will flow from that decision that we make that will have consequences that will be fairly significant.

My concern is that it is worth taking a step

back, in my view, and having the working group, the taskforce, look at this in detail. Look at the legal implications. Look at the implications that people have addressed in other documents.

While we may agree sitting around the table, it is such an important decision that it is worth having the working group look at it in great detail, look at the implications, and then bring it to the committee in some format with better documentation so that we can make an informed decision about the implications of these kinds of central questions.

I'm concerned that just sitting around the table and talking will not get at some of these concerns.

DR. EVANS: I understand what you are saying. What I'm trying to advocate, though, is if there are certain subjects that we all do agree on, in a way I'm not sure whether all of the implications, mapping those out, and spending three months doing that, is worthwhile.

I think that there are certain aspects to DTC that rise to the level of obviousness, such as BRCA testing as a medical test. We don't need to spend three months figuring out the implications. I'm just putting this out

there. It might be worth highlighting those things that we all agree rise to importance without spending months and months more.

DR. WISE: Basically, by doing that, you will be articulating a little more clearly what the question is, but you are not going to be providing much guidance on how to deal with it. If we are talking about what is included in your box, how do we identify which are clearly medical tests and which may be medical tests and which are recreational.

My concern is that we do this right. The implications here not only speak to the DTC community but also to the utility and legitimacy of this group. We have a really great report. It took quite a bit of time and thinking to get this through.

My concern is that by sitting around the table in a short amount of time we are going to completely overshadow anything contained in this report that was considered over a few months with a decision that we are taking without a more formal and more deliberate process for making decisions that are going to ripple through the whole conversation later on.

I'm not a big fan of waiting three months. I'm not a big fan of waiting for anything, in general. I just think that this is an important decision that is going to have implications, as we heard, for a variety of agencies. We need to do this right. Members of the committee that might not be directly involved day-to-day with DTC kinds of issues need to have background information that has been vetted and articulated well so that we can make good decisions about these kinds of issues.

DR. EVANS: What I would say is I want to do it right, too. The decision, then, around the table I would phrase as, are there issues that we all can agree on that don't need months more of deliberation or are there not. If there are not and if we are happy with this report, then so be it, we go ahead with this report, or perhaps we delay it and do some more things.

Again, I just want to throw out for the consideration of the committee, are there some things that rise to the level where we might want to say to the Secretary we have concerns about XYZ. I would throw out there that emphasizing that there is a need to reconcile claims with reality does rise to that level, but I'm just

one member of the committee. I think we should discuss that.

DR. WISE: We have to say more than just that these are concerns. The Secretary already knows what the concerns are.

DR. EVANS: No, no, she doesn't. She does not.

DR. TEUTSCH: What I'm taking away from this conversation is these tests have not necessarily been considered medical tests. It is a significant change for this committee to say that they are medical tests when they deal with those medical issues and they need to have the same kind of oversight that you would for other types of medical information.

Now, that is the core. If we can get there today and get some agreement, we can get it back and put this in a page or two. We can then highlight some of the other things that we have done that need to be brought to bear on this. Highlight some of the other issues, but keep it fairly focused.

This would be a substantial change and contribution, and doesn't really require a lot more research, if you will, for us to make the statement that

they should be considered in that context.

MS. AU: This would narrow the medical tests. We would explain what we are talking about.

DR. TEUTSCH: This is a set of tests that are being offered directly to consumers. Those that Jim just described, that is what we are talking about here.

DR. GUTIERREZ: Perhaps a little history would help here. About two years ago, when the genomic scans began to come onto the market -- and this is public. The companies actually talked about this -- they came in and spoke with the FDA because the FDA wanted to know what kind of claims they were making. Invariably, most of them were telling us that the claims they were making were not medical claims.

Things have changed since then. I do want to note that the claims seem to have changed and the types of tests have changed, but it is on the record that they claimed that these are not medical tests.

DR. BILLINGS: Do we have consensus, then, about what a medical test is?

DR. EVANS: I don't think we need consensus about the general definition. What we need consensus on is, are

they performing some medical tests. I think the answer to that is obviously yes. They are doing BRCA testing and LRRK2 testing, period.

DR. BILLINGS: I can see us saying that we want one standard for medical testing, but I think we also then need to be clear about, is there some other kind of testing besides medical testing and what is that.

DR. EVANS: Yes. If you want, we could give examples. We could say ancestry testing is not medical testing. We are not endeavoring to define the entire landscape of medical testing, but it is like Justice Potter Stewart said, I know it when I see it. BRCA and LRRK2 is medical testing.

MS. AU: I have Marc and Phyllis, and we have one minute.

DR. WILLIAMS: I think we need to move forward. We have discussed the medical test issue in the context of the Oversight report. I don't think we need any additional work on that. I feel comfortable moving forward to say we need to have one standard and these companies are performing within their suite of tests some tests that are clearly medical.

DR. FROSST: I want to address the point that I think that there might be some confusion on. That is, when we talk about DTC, we talk about a very, very big range of genetic tests offered directly to the consumer without a health provider, right? That is an enormous arena.

What I think some of us are more specifically talking about are the types of genome scans that are being done by 23andMe, Navi, et cetera. I think these are two overlapping but not necessarily different arenas. There may be some discomfort in making a broad statement like "You know it when you see it" about what is medical.

DR. EVANS: The point is that these whole-genome scans, I agree, contain many different things. Some of them are clearly medical.

DR. FROSST: I totally, completely, 100 percent agree with you. If we are talking about specifically that realm of tests, then we need to specifically say in terms of whole-genome scans that contain things which are medical that this is what we are talking about, rather than Bob testing for six things in his garage and recommending vitamins.

DR. EVANS: It is like Steve said. It is the

subset of tests within these suites that rise to a level by which one would call them medical testing.

DR. FROSST: Agreed.

DR. LICINIO: I just have one comment. I would add "medical and behavioral." They could say someone has a gene for bipolar and that is not medical, it is behavioral. So I would put "medical and behavioral."

DR. TEUTSCH: Health-related.

MS. WALCOFF: I want to get to the next step of that. If we are making this broad statement, what does that mean. I think Paul was getting to that a little bit more. Are we really saying there should be a single standard or that these tests should be held to the standard of? I don't think it is as helpful to basically just call them out and say, everyone knows you are making a medical claim and you are saying you are not. I think we should say something that is actionable by FDA or CMS.

DR. FERREIRA-GONZALEZ: If we say these are for medical purposes, we have the whole report on oversight.

DR. TEUTSCH: Let me just get a straw poll from all of the folks here. I think we have gotten to a core set of issues that we have just articulated. These are

health-related tests. They should adhere to the same standards as they would if they were being used in a clinical setting. We can work on a relatively short document of a page or two that is going to highlight that and refer back to what we mean when we say there is oversight. We have these other reports that will be in the attachments.

I think it is important because a humongous amount of work went into getting this to this point based on what we thought the last time. I think we have come a long way in this discussion. It has been a very constructive discussion, but I would like to get some agreement from this committee that you are comfortable.

If we go back and bring something to this group in October, is there a general consensus? Can I just take a straw poll? How many conceptually are on the same page with that?

DR. WILLIAMS: I'm sorry?

DR. TEUTSCH: With a two-page report that basically says that when they are health-related tests, they contain medical and relevant information, that they should then have the same type of oversight as those that

would be used in a medical environment.

DR. WILLIAMS: We wouldn't look at that until October?

DR. TEUTSCH: You will get a chance to see it in October.

DR. WILLIAMS: But it will go out before then?

DR. TEUTSCH: No, no. We will bring it back for approval by this committee. We will spend the next three or four months getting it in shape.

What I don't want is to bring that back and have people say, I don't agree that these are medical tests. I would like to make sure we are on the same page.

MS. AU: That would give me a chance to schedule that 4:00 a.m. conference call.

MS. WALCOFF: So we are going to say if they make health claims, they should be held to the same standard as other genetic tests that make health claims?

DR. TEUTSCH: Other tests.

MS. WALCOFF: Other tests that make health claims.

DR. LICINIO: They may not be making those claims, but if they test for things that are medically

relevant --

DR. TEUTSCH: Providing health information.

MS. AU: We will have the taskforce come up with the definition.

DR. TEUTSCH: Yes, we will get to the wordsmithing, but that is the point.

MS. WALCOFF: It sounds like it is a combination of what you raised earlier with basically created a focused executive summary.

DR. TEUTSCH: Exactly.

DR. EVANS: We have to address the reality, not just their clients.

MS. WALCOFF: I will agree to making a focused executive summary. I'm happy to help, too, since I was a latecomer and adding more work. I'm not the only one.

DR. TEUTSCH: No, no. I think this has been an excellent discussion.

MS. WALCOFF: Also, it is unfortunate Barry is not here this morning for this because it would be interesting to get some feedback from him as well. Maybe we can circle back with him.

MS. AU: Barry?

MS. WALCOFF: Barry Straube from CMS. They are obviously heavily looking at this area as well.

DR. TEUTSCH: We dealt with a lot of those issues in the Oversight report.

Is there anybody who has a problem with that general approach? You will see it again. You will have a chance to discuss it.

DR. WILLIAMS: Steve, I don't have a problem. I agree with the approach. Does it need to also focus on privacy and security in addition to that? Will just calling these clinical medical tests give us enough framework to talk about those issues?

Yesterday's discussion by our group was almost exclusively focused on that. When someone came forward with a very different presentation, we all leaped to those very great concerns.

MS. AU: There are a lot of other issues that depend on how the testing is done and that have nothing to do with whether they are health-related or not.

DR. TEUTSCH: We will need to get some of this back to a committee to work on because we have heard a bunch of other issues. I think what we have heard is that

the oversight protections and those kinds of things should be the same as in the medical arena.

MS. AU: HIPAA might not work.

DR. TEUTSCH: No, but that is what we need, new policies.

MS. AU: Do you want to expand that portion? Are we expanding the report at all with some of the other concerns?

DR. NUSSBAUM: I'm just trying to figure out whether there is one overarching theme, that these are medical tests, or whether there are two or three subthemes that people are concerned about. It doesn't change, I don't think, the significant work that has been done that is the key statement. I just didn't know if we wanted to include that.

DR. TEUTSCH: I think it is implicit. We will need to work those things through because basically we are saying they are medical, they are not just recreational or curiosity.

DR. FERREIRA-GONZALEZ: I think we might have to defer these issues. If we say that these tests are medical tests, HIPAA comes into play.

DR. TEUTSCH: Exactly. Those are the protections I think you are referring to.

DR. FERREIRA-GONZALEZ: That is what I'm thinking. This idea of selling the data, there is at least a subset of information on that.

DR. NUSSBAUM: Clinical validity, HIPAA, everything else just naturally follows.

DR. TEUTSCH: You are probably right. We need to be able to indicate what are the things that follow from that recommendation.

MS. WALCOFF: Right. That is well articulated.

DR. FOMOUS: To go back to Sylvia's question, are you wanting us to add, in essence, new recommendations? The paper does discuss the problem with HIPAA. These companies are not a covered entity under HIPAA, so HIPAA won't apply to them. Are you asking or suggesting that we should also include for October new draft recommendations that these entities should be covered under HIPAA? That is just an example.

So the question is, between now and October are you also asking the taskforce to come up with new recommendations in addition to recycling some of the old,

or do you just want to go with the paper that we have with the preface or the executive summary in front of it addressing the medical test issue?

DR. TEUTSCH: I want to make sure we have no dissent on the substance on this. Then I think we have to take it back and really look to make sure that the appendices are germane. We can do that as staff work.

We have to move on. Are there substantive problems with the general approach or the general statements that we have made?

DR. FOMOUS: I just want to clarify the scope. We are not going to do new stuff.

MS. AU: No new recommendations.

DR. FOMOUS: No new recommendations. We are just going to fix what we have.

DR. TEUTSCH: This is fundamentally a recommendation about this is a medical test.

DR. EVANS: In the deliberations of the taskforce over the next few months, if it came up that we should have a bullet about privacy, we could come back to the committee with that, too, right? So it is not that we would be off limits from considering any of those things where we had

concerns that we thought might not have been adequately addressed by prior recommendations.

DR. WISE: The committee is asking you to go back and make a recommendation around this medical testing issue.

DR. FOMOUS: Right. I got that.

DR. WISE: That is not a recommendation here. Therefore, it means deliberation in the group, more work, and bringing it back in three months for consideration and approval by the committee.

DR. TEUTSCH: This has been great, and very helpful. Actually, the committee has done a huge amount of work in a very short period of time that I think is going to move this all forward. I think we will be able to build on and use what you have already done. We will bring it back here for lively discussion the next time.

MR. BOWEN: Steve, could I make a quick announcement related to DTC? Several folks here were involved in a workshop with CDC and NIH in December on the scientific foundations of personal genomics. Those recommendations will be published in Genetics and Medicine in September.

Also, CDC looked at DTC perceptions and use among consumers and physicians in the Doc Styles and Health Styles survey in 2008. Those results will be published in Genetics and Medicine in August. I just want folks to know about that.