

Overview of Report, Summary of Public Comments on Draft Report and Goals of Session
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DR. TUCKSON: Welcome back. I want to make a special note and a special hello to the 32 people who are so committed to what we are doing that they are listening. Even though the video is not on live, there are 32 people listening live to the audio. You are beyond terrific, and we think you are wonderful.

You need to know that the video part is actually in a delayed broadcast so that it will be up in a couple of hours and people will be able to watch it on a time delay deal. We are told that tomorrow the live video will be up in the morning. There is a video to it, but it is just going to be time-delayed. So we are appreciative of all of that.

Now to the discussion. We have heard a lot. As we go into the discussion, it is critical that we have a lot of stakeholders who have voices in this. I thought [Sharon] was very passionate about thinking through and being balanced.

There is also this issue of the chilling effect of over-action and the harm of too much action that people have been equally passionate about. So we will work through these deliberatively. Thank God we have Andrea to lead us through it. Andrea, I will help be a traffic cop, but the floor is yours.

DR. FERREIRA-GONZALEZ: Thank you, Reed. Can everybody hear me in the back. Great.

What we are going to do now is go into an overview and the goals of the oversight session. Before we dive into specific discussions and specific recommendations, I'm going to give you a little bit of a background of when did we receive the charge and what actually occurred until the point that we are to date.

As Reed mentioned, most of the meeting is devoted to the discussion and deliberations we need to have as a full Committee on the draft recommendations that have been developed to address the Secretary's charge to us on the oversight of genetic testing.

Before we begin the process, I'm going to review our charge, the process we used to draft our report, the public comments we received on the report, some of which were just reiterated for us, and the changes we have made to the report in light of the public comments and further deliberations.

Although I'm going to be reviewing comments that pertain to both the recommendations and the report, I want to emphasize that we won't be discussing the report at this time in great detail.

The focus of our deliberations in the next two days will be to finish with the recommendations. By the end of the session tomorrow we hope to have consensus on the recommendations and approval of their transmission to the Secretary. We will also seek the Committee's approval on principle. Keep that in mind. We just want the approval on the principle of the document. That will be further, later, edited through the month of February and then submitted at the end of April to the Secretary.

I want to take a moment to highlight the Secretary's charge, which we received at our March 2007 meeting, to remind you that our final report and recommendations should address the issues that were raised by the Secretary in the charge. It begins with a request to develop a comprehensive

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map of the steps needed for evidence development and oversight of genetics and genomic tests, with the improvement of the health quality as a primary goal.

I think we need to emphasize some of these issues that we have responsive to the charge of the Secretary, and we actually have done that.

The charge also tasked the Committee to evaluate existing pathways that examine analytical validity, clinical validity, and clinical utility, attributable harms if these pathways are inadequate, and the roles and responsibilities of the relevant government agencies and private sector organizations. We were also asked to consider whether genetic tests are different from other laboratory [tests] for oversight purposes.

Additionally, the charge asked several questions about proficiency testing: communication pathways to guide the use of genetic testing and new approaches or models involving the public and private sectors to demonstrate clinical validity and develop clinical utility.

Lastly, we consider whether additional or revised government oversight would add value to our patients.

As we received the charge from the Secretary, six members of the SACHS Committee volunteered to be part of this group. We needed a mass to be able to deal with this issue, formally called a steering committee. This steering committee actually took the lead in looking specifically at the charge and, from the charge, developing the scope of what our document was going to be.

As we looked through the charge and the scope of what we actually intended to respond to the Secretary, we started devising how this document was going to be organized and divide it, actually, into different chapters.

Ad hoc members and field experts were brought in as we started to realize the scope of the charge. We needed additional individuals with different types of expertise and also federal experts, and I have listed all of them here.

I would like to thank the steering committee and the ad hoc and federal experts that devoted a significant amount of time from their personal time to look at these issues. We had a number of different meetings, conference calls, two face-to-face meetings in July and September, and have worked tirelessly to actually come up with the document that you see today.

We have discussed the activities of the taskforce in detail at previous meetings, so I'm not going to go into detail. What happened throughout the different times is just listed here. This slide only briefly summarizes those activities.

We began drafting sections in the report in May, and we were divided in different chapters. The reason we divided into different chapters is just to be able to tackle some of the incredible tasks that we had in front of us.

We worked very frantically through the summer and the fall, and we were able to, with a conference call with the entire SACGHS Committee, put out a draft report for comments. Those comments took place from November 5th to December 21st.

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During the public comment period, we received 64 sets of comments from a range of different stakeholders. I have listed here the different stakeholders that we have received information from. You also have a summary of these comments in Tab 3 of your briefing book.

As you can see from this slide, the majority of the comments were from professional organizations. Some of them we have already heard this morning. Twelve from industry, 11 from government agencies, five from healthcare professionals, six from advocacy organizations, four from academicians, and one from one individual.

The steering group began analysis of the comments in late December. Actually, we received the packet and we were so excited to be doing this over the holidays, but I really want to thank the whole steering committee for actually taking the time over the holidays and the entire [month of] January to go through the exhaustive review of all the public comments.

We met by conference call the first three weeks in January. Every Wednesday afternoon we had a conference call, and we discussed the public comments specifically and what changes or revisions need to be made to the recommendations and even the text.

The revised recommendations were sent to the taskforce. Again, we are getting into the taskforce to get a wider perspective from individuals that had already contributed. We had a conference call on January 23rd.

In addition, we invited taskforce members to present additional comments, and we will have a member of the taskforce actually present later today.

After that January 23rd [meeting], we actually made some changes to the recommendations, responding to some of the comments and discussions that we had with the taskforce and presented this to the whole Committee.

You will remember our conference call on January 30th. That conference call was not to start discussing the recommendations but just give you an idea where we are and for us to get a sense of where are the areas that might have some further discussions or deliberations that are needed today and tomorrow.

Although most of the comments are for edits or comments for specific sections for the report, the overall tenor of the comments was very positive. Most commenters thought that the report was responsive to the Secretary's charge and provided an excellent review of the issues associated with the oversight of genetic testing. Commenters also recognized that the development of the report involved diverse stakeholders.

Some recurring themes that also emerged from the public comments are listed in this slide. Several commenters were concerned that the report's broad definition of genetic test may slip non-genetic testing under this report. To address this concern, actually we have made changes to the text to provide further examples of what are actually considered to be a genetic or genomic test.

Many commenters agreed with the report's stand on genetic exceptionalism. For that oversight purpose, genetic tests are not different from any laboratory test. They may defer in other ways, however, such as communication of the results to the patients or even healthcare professionals.

I think it is important to note that there was strong support for the increased proficiency testing or genetic test and development of standards and reference materials needed for proficiency testing. A large number of the commenters were very positive about these developments.

Comments for a registry of genetic tests favor a mandatory approach, but there was no clear indication where such a registry should [be housed.] A few comments articulated preference for CMS or FDA, but in reality most of the commenters did not say anything about the registry. So those that did actually make some comments, which was not the majority, did provide different places where these registries should reside.

Commenters also had concerns about direct-to-consumer advertisers and consumer-initiated testing and agreed with the recommendation to improve enforcement of the current regulations to cover this type of testing.

In addition, there was overall agreement that enhanced oversight is needed for genetic testing. FDA's authority to regulate laboratory tests was not questioned. Its risk-based regulatory approach was affirmed, although there were some comments criticizing specifically the FDA's IVDMIA draft guidance.

Commenters recognized that there is an adequate evidence for the clinical validity of many genetic tests and the importance of adding to these two evidence bases. Although many agree with FDA roles in assessing clinical validity, some commenters asserted and favor CMS's role in this particular issue.

In addition to addressing gaps in clinical validity, commenters also called for more evidence and analysis of clinical utility and increased education efforts to enhance genetic knowledge of healthcare providers, public health officials, consumers, regulatory officials, and actually payers.

Last but not least, before increasing oversight, commenters asked that benefits and harms to patient access and cost should be considered. I think we looked at that and modified some of our recommendations in particular to those comments to make sure that the stakeholders are part of any of the recommendations that we are putting forward.

We made some revisions to the report as a result of these public comments and further deliberations not only on the steering committee but also with the taskforce, and we added public health surveillance as a key consideration in the executive summary, added an introductory paragraph explaining transient genetic tests, added a methodology section to explain reports development -- how did we get to the development of the report -- and also revised the definition of genetic test to include genomic tests and examples of the tests excluded from the definition. We are hoping to be a lot more clear about that.

There were also other revisions of the report where we added information about the Senate Bill 1858, added the role of the states in oversight of newborn screening, added activities of the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children to roles of federal agencies in research and development and evidence, which is located in Chapter 2 under "Knowledge Generation."

We augmented the discussions in nanotechnology to include devices using extremely small amounts of materials. We added the term "reproducibility" as a key term in Chapter 4. We updated and actually corrected information about the CAP products and PT performance. We corrected information about transport of biological materials and actually augmented the list of

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professional societies in Chapter 4 to make sure there are as many as we can list. But again, we can't do an exhaustive listing of all the professional organizations.

We have also added other activities that are currently underway at ACHDGDNC and HRSA to discussions of the clinical utility of the test and patient access to genetics expertise.

We added a discussion of harms due to inadequate information about clinical utility. We corrected information about Oncotype-DX. We had actually mistakenly called it FDA-approved when it is actually a laboratory-developed test. We have updated statistics for board-certified genetics M.D.s, laboratory disciplines, and genetic counselors.

We added information about privacy concerns related to direct-to-consumer testing and advertising and commercially operated PHARs.

We added ACMG-AAP-developed ACT sheets and algorithms as examples of critical decision support tools that are currently being used for newborn screening.

The next steps. Here we go to today. As you can see, at the end of February the final recommendations and the revised draft report will be submitted to the Secretary. Today we need to further discuss the recommendations and actually come, tomorrow afternoon, to finalize the recommendations that will be submitted to the Secretary.

We will still be receiving comments on the draft report even though we will not be discussing the draft report. We will ask you to approve it or not in principle, but the edits will be accepted until February 20th to make sure there is enough time to continue the editing of that draft report.

So, the final recommendations and revised draft report [are to be] submitted to the Office of the Secretary by February 29th. Then we will continue to work on the report over March until April 16, where we will have a final review by this entire Committee, maybe through teleconference.

April 30th [is when] the final report will be formally submitted to the Secretary.

So again, just to make sure we all understand and are on the same page, our focus is to finalize recommendations by the end of the meeting. Edits to the report content can be sent to Cathy. I think we all [know] Cathy Fomous has been incredible in support to our efforts here.

[Applause.]