24th Meeting of The Secretary's

Advisory Committee on

Heritable Disorders in Newborns and Children

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DR. HOWELL: Well, first let me wish everybody a very happy Cinco de Mayo. And having gotten that greeting out, let me welcome you to the 24th Meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. This committee has been an extraordinarily productive committee, et cetera. And the work that's come out of it is some work that I'm extremely proud of.

And today, it's my privilege to welcome three strong, new members of the committee. And this makes me encouraged that the work of the committee will go forth stronger than ever.

The first I would like to introduce is Dr. Donald Bailey, who is a distinguished fellow at the RTI International and the Research Triangle in North Carolina. Dr. Bailey's research addresses the early identification and early intervention for children with disabilities as well as family adaptation to disability. Currently, Dr. Bailey directs the NIH and CDC-funded projects focusing on the Fragile X syndrome and broader issues surrounding the ethical, legal and social consequences of genetic discoveries.

He has recently completed a two-year term as President of the Board of Directors of the National Fragile X Foundation.

And, Don, we're delighted to have you join the committee.

DR. BAILEY: Thank you.

DR. HOWELL: The next person to introduce is Dr. Fred Lorey, right over here. Fred has just arrived after a long flight from Los Angeles. And Fred has been a member of the California Department of Public Health and Genetic Z Screening since 1994. And he currently serves as Acting Director of the Department of the Newborn Screening Program in Richmond, California, which I think, as everybody knows, is one of the largest newborn screening programs in the world and certainly the largest individual program in North America.

Fred is extremely familiar in this committee because his efforts and his department have been extremely important in a lot of the major pilot studies that have been done in newborn screening as well as other investigative aspects of newborn screening.

And, Fred, we're delighted to have you formally join the committee. You've been working for the committee since, I think, the committee started. And welcome very much.

And the third person of this committee is Dr. Alexis Thompson, who's sitting between Jerry and Jeff over here. And Dr. Thompson is currently the Medical and Scientific Director of Hematology at Children's Memorial Hospital and associate professor of pediatrics at Northwestern in its Feinberg School of Medicine in Chicago. Her clinical interests include the hemoglobinopathies, bone marrow failure syndrome,

stem cell transplantation. And her laboratory research is focused on developmentally regulated genes in early hematopoieses.

And welcome, Dr. Thompson. And your area of expertise will be valuable as we go with these various recommendations in the coming month.

Now, I have a note that Ms. Harris has some housekeeping notes. And all we have to do is find Ms. Harris. I guess we're not going to have any housekeeping today. Hopefully, you will do your own dusting and so forth.

First, each of the members have the meeting summary for January 10th. And I would like to hear, are there any changes or objections to the minutes, as you've received them? Hearing no, I will need a motion from the committee to accept these minutes. Can I have a motion?

MALE SPEAKER: So moved.

DR. HOWELL: Second?

MALE SPEAKER: Second.

DR. HOWELL: Those favoring the adoption, raise your hand. Any objections to that? That appears to be unanimous.

We have a lot of committee business to discuss this morning. Some of the correspondence that we're going to be discussing it

will be of great interest to the committee. But I also know that it's of great interest to a number of people in the audience. During the committee discussion, we will not have audience comments about the correspondence. But we will look forward to hearing audience comments during the open discussion later this morning.

Under tab 5 in your briefing book or in your laptop are the two letters from the Secretary, one regarding our residual blood spot recommendation. And the severe congenital heart disease response letter is also located in your briefing book. And you also have that on the thumb drive.

Let me first draw your attention to the letter concerning both the residual blood spots and the congenital heart disease issue. The Secretary has referred both of these recommendations to the newly-formed HHS Interagency Coordinating Committee, which has not yet met, but is anticipated to meet very soon -- and that's called the ICC -- for their review and input. The ICC is comprised of the National Institutes of Health, the Centers for Disease Control and Prevention and Health Resources and Services Administration, the Agency of for Health Care Research and Quality and the FDA.

In her response concerning the residual blood spot recommendation, the Secretary states that, "The use of the ICC will allow

for more formal engagement of the Office of Human Research Protection and the Office of Civil Rights along with the federal agencies assigned to the ICC for its authorizing legislation." And I think that this group is fully aware of the fact that there's been tremendous interest in the whole issue surrounding residual blood spots. There's been a lot of programs and so forth.

And I might also call your attention to the fact that the committee's report entitled, "Consideration and Recommendation for the National Guidance Regarding Retention and Use of Residual Dried Blood Spot Specimens after Newborn Screening," has been accepted for publication and actually is in press in Genetics in Medicine. It will be published in two ways: an abbreviated form, substantial abbreviated form that includes all of the recommendations of this committee. But then on the online version, will be the complete report, which is much more expanded. And the authors of this is the working group of this committee. And you've all seen this report.

But I think that this will create a great deal of interest. And there has been an enormous amount of interest in guidance from this committee about the storing and use of residual dried blood spots. And I think this report summarizes that nicely.

Would anyone like to comment about the residual blood spot

issue? I just was made aware that this has been accepted for publication yesterday. And I doubt that this is still in galley proof. But as soon as it's in its final thing, it'll obviously be distributed to this committee and will soon be available. It'll be published in the June issue of Genetics in Medicine. Any comments about that? We'll look forward to that. And hopefully, there will be some helpful recommendations that will come from the Office of Human Research Protection and the Office of Civil Rights. Both of those groups, obviously, have a keen interest in this.

In the Secretary's reply to the critical congenital heart disease screening recommendation, the Secretary noted that our letter to the Secretary discussed recognizable evidence gaps regarding screening for critical congenital cyanotic heart disease. And her letter goes on to explain that, after consulting the HHS leadership, she determined that the advisory committee's recommendation were not ready for adoption. Because she acknowledged that this is such an important issue, she has, again, referred this recommendation to this coordinating committee. And this committee has not yet met, but their first agenda item would be the congenital heart disease.

The ICC leadership has actually examined the evidence gap described by the advisory committee and proposed a plan of action to address the identification of effective screening technologies, development

of diagnostic processes and protocol, education for matters in the public and the strengthening of service infrastructure. She has also directed this committee to respond within 90 days, at the latest, and to keep the committee informed. And your thumb drive also contains correspondence of the Secretary expressing support for the committee's recommendation.

We've had an enormous correspondence about the report from this committee about screening for congenital cyanotic heart disease. And those letters, I think, are all there in your thing. Now, I think that this committee is very much aware and you've been very much on top of it. But after our recommendation to the Secretary that this be included, it's traditional then to start looking at the implications of such a recommendation.

And this committee convened a meeting at the Heart House here in Washington that included the American Heart Association, the American College of Cardiology, the American Academy of Pediatrics, large numbers of people from the public health sector, from neonatology and met in an extremely productive session that spanned a couple of days that laid out detailed plans and also looked at, again, the information about screening technologies and so forth. That also included the folks from the U.K. that had finished a document on newborn screening. They were here and presented all their information.

And we were privileged to get that, actually, before it was published. The U.K. permitted us to look at that. It has since been published. But we also had the folks from Scandinavia that's had the considerable experience in that area.

And this group, this working group, has developed a document about implementing newborn screening for severe congenital heart disease. And it's been very interesting to see the very positive response to that.

And I'd like Tim Geleske just to talk about one positive response we've had that was extremely prompt and very much appreciated.

Tim?

DR. GELESKE: Thanks, Rod. So the work group report from the critical congenital heart disease work group went to the academy for review. It was reviewed by various sections and committees. And based on their comments, the executive committee of the Board of the American Academy of Pediatrics has endorsed the work group's report. So I believe next steps will be that it will be published in the Journal of Pediatrics along with a statement confirming that endorsement.

DR. HOWELL: It's rather unusual to have such a quick and brisk response from such a large organization supporting this. And it's

pointed out, Alex Kemper is currently working on the article that will be in Pediatrics that outlines that.

Do we have any comments -- and I'm going to have a good bit to say about this, I might point out. But let me hear from the committee before you get to my comments.

DR. BUCKLEY: So, Rod, what does it mean if our ICC committee comes back and says that they do not recommend approval? Can we go forward or not?

DR. HOWELL: Well, I think that this committee has made a recommendation to the Secretary. And if this coordinating committee makes a recommendation that it not go forward, I would assume that the Secretary will take their advice and not recommend that it be included in the panel. That would not mean that our material that we've sent forward and that's been published would obviously go away.

DR. BUCKLEY: And that would not preclude states from doing this on their own, I guess?

DR. HOWELL: One of the things that's happening is that there is -- and we'll hear about some of this in the public commentary area because this has been such a clear recommendation by many assessments that states are proceeding on their own. For example, there's been a bill in New Jersey that's been passed by both the House

and the Senate there to proceed with newborn screening.

DR. BUCKLEY: I saw that.

DR. HOWELL: And I would assume that, based on these recommendations and also the recommendations of the professional groups, that individuals and states will probably go ahead with that.

Alan?

DR. FLEISCHMAN: This may sound like a silly question. But the materials that were sent to the Secretary and then transmitted to the ICC are one point in time, and a tremendous amount of effort and more evidence has been amassed since then. So is there a formal mechanism for that learned group to receive, in a formal way, the evidence and the additional materials?

DR. HOWELL: I don't know how this group will meet, since they've never met. But maybe Michele can comment about the logistics of that. I don't know the answer to that about what is the mechanism by which this group will have any discussions with this committee or anything of that nature.

Michele?

DR. LLOYD-PURYEAR: Coleen?

DR. HOWELL: Coleen? I think Coleen has been involved in the formation of this committee.

DR. BOYLE: So, actually, just a little update. The ICC did meet yesterday.

DR. HOWELL: Oh, good.

DR. BOYLE: So, and it's co-chaired jointly by CDC and HRSA. And Bia Strickland is chairing it from HRSA perspective and Carla Cuthbert, from CDC. So we did meet yesterday and talk about moving forward on our task from the Secretary. And that really is specific to the recommendations in the letter, which was to look at the five recommendations and, sort of, deliberate as to how agencies can respond to those specific recommendations that the committee had made back in, whenever it was, in September.

DR. HOWELL: And the committee, I would assume, will obviously have all the activities of the working group that met at Heart House?

DR. BOYLE: Correct. Correct.

DR. HOWELL: Which was a big opportunity.

DR. BOYLE: And the charge is to get back to the Secretary within, I think it's, 90 days. Is that what it was?

DR. HOWELL: The charge is within 90 days. Now, is there a mechanism for this group to get specific information or have further meetings with this committee?

DR. BOYLE: That's not the plan. The plan is to go back directly to the Secretary.

DR. HOWELL: Okay.

Fred, you had a question or comment?

DR. CHEN: That answers my questions, thank you.

DR. HOWELL: I took it upon myself to call Dr. Mary Wakefield, who is the administrator of HRSA and its oversight of this group. And I called Dr. Wakefield about two issues that were of concern to me. I outlined the mechanism by which this recommendation had been made with the evidence that had been gleaned and also the information that had come from the Heart House group and the experts and so forth and the fact that it had been so generously endorsed by the groups that had convened and simply expressed the fact that I was concerned about the recommendation. Obviously, that was a personal opinion, but that was of concern to me.

And I also was concerned that delay is a big issue. This is such a common problem. And the information that we saw at Heart House would suggest that as many as four or five infants per day might benefit from this screening test. And so, that when you start talking about a month or three months, to me, that's important. And that's a personal opinion. I'm very sensitive to that issue and so forth.

Dr. Wakefield explained again the position of the Secretary, who she obviously represents and was very courteous, et cetera. And we had a very pleasant discussion that she was very -- I was very crisp about my opinion of the subject.

The second area I wanted to comment about is that while I was on vacation on April 21st enjoying myself in Miami with my family, I got an e-mail at 1:30. Actually, it was 1:32. I wrote it down here in my notes, that there would be a -- on my BlackBerry -- that there would be a conference call of Dr. Wakefield and other leaders of CDC and HRSA and the NIH to discuss this recommendation. And I found that quite surprising. I have not yet seen the letter of the Secretary. But it announced that this meeting would be at three o'clock. This e-mail came at 1:32.

But I was fortunate enough to get an e-mail copy of the Secretary's letter at 2:45. So I did have it before the meeting. It's my impression that none of the appointed members of this committee were able to get that call because everybody's busy and were committed. And I thought that was very short notice of an important thing.

And Dr. Wakefield advised me that the plan was that that was really for the stakeholders that had corresponded with her, et cetera. I also was concerned in that I was unable to make a comment. Although there was instructions that if you wanted to make a comment, you were

supposed to push star, one or something of that nature. And I pushed star, one, as did others. And unfortunately, our star, one led nowhere.

But anyway, apparently this conference call was arranged by a contractor. And Dr. Wakefield was very courteous and apologizing on the short notice, et cetera.

But are there other comments from the committee about this? And we'll try to move on.

DR. VOCKLEY: Yeah, Rod, just a question. Trying to keep my remarks to sounding crisp as opposed to anything stronger, is there a way for this committee to officially express concern about this mechanism of review of the review of the review of the evidence? We have a process -- I mean, this committee is the expert group in this area. When you make a recommendation, it's obviously the Secretary's prerogative to accept it or not. But to say we're going to review the evidence again seems somehow to negate the relevance of this group. And I don't want to do all of this work for nothing. And I think that there ought to be some sort of carefully-worded and therefore, not written by me, response of the ICC back to the Secretary that just expresses some official concern as opposed to your personal concern about this mechanism for reviewing committee decisions.

DR. HOWELL: I think that the committee, the ICC is

established in law. And I think that it's very worthwhile to have an oversight committee that looks at all major recommendations coming to the Secretary.

But I think Peter might have some specific comments about -- and also --

DR. VAN DYCK: Well, just to clear up any misconceptions, the Secretary is very appreciative of the committee and the work of the committee and looks to the committee for expertise, scientific and medical expertise. She recognizes the hard work the committee has put in. As Rod announced when he did his introduction, this is the 24th meeting. This is an extremely productive committee.

But the letter itself mentioned gaps in evidence. And the work group report itself mentions gaps in evidence. And the gaps are around implementation. So this is not second-guessing the work of the committee. It's trying to address the gaps in evidence that are the responsibility of the agencies to implement.

So it's going beyond the scientific and technical expertise of the committee and trying, for the agencies that are necessary to be onboard and to implement this recommendation in the most sound way, to get together and analyze how they can best do this and report back to the Secretary in really a very short timeframe, 90 days. And the committee

has already met, as you've heard, yesterday. And this is a high priority.

Ninety days, we can't go beyond that. But we can complete our work sooner than that. And that recommendation then goes to the Secretary. So it is addressing the gaps in evidence that were mentioned in your letter, specifically, in the committee's, our, letter, specifically and not questioning the scientific and medical expertise of the committee itself. So I don't know. The agency heads, Dr. Frieden, Dr. Wakefield and Dr. Collins, all wanted some additional analysis of the gaps in evidence and recommendations for their agencies. And this, then, is set up by law as their way of getting that information analyzed and to the Secretary.

And I don't know if Coleen or Melissa want to say anything in addition, from NIH or CDC.

DR. HOWELL: Let me make one comment before. I should have introduced Melissa Parisi, who is here today as an alternate for Alan Guttmacher, who was unable to be here. And Melissa is his official alternate. And we welcome Melissa today.

DR. PARISI: Thank you. If I could just echo Dr. van Dyck's comments. I think the perspective from NIH as well is one of the charge to the ICC is really to address the issues with regard to implementation of the recommendations from this committee. And that's really the focus. And the ICC is taking this very seriously and does have a very tight framework

with which to do this, the tight timeline, and will do its best to respond appropriately.

DR. HOWELL: Jeff?

DR. BOTKIN: The recommendation the committee made about congenital heart disease, sort of, wasn't straightforward. We did have a couple of caveats with that. So it seems to me that this is an opportunity for us to think about other circumstances in which similar sorts of recommendations will be made, if, in fact, it's not simply -- accept screening, but rather accept, given some contingencies here. And those did involve potentially additional research to fill those gaps.

I guess, the question then comes back to our committee, how should we deal with those types of contingencies with future votes. Do we need to think those through so that it doesn't end up with a review of the review of the review? At the ICC level, are there ways that we can address those in an efficient manner of coming out of the committee so that we don't get into this, sort of, challenge in the future. Because presumably, we're going to confront other screening modalities where we think it's appropriate to go forward, but yet, strongly feel there's additional data that need to be collected.

DR. VAN DYCK: And I might add one other thing I forgot to say was, it is not the presumption that the ICC is set up to review every

recommendation coming from the committee. It is only those that are necessary for additional information, that may be necessary for the agencies' involvement in implementing the recommendation. So it's not an expectation that there's a second review of the committee's recommendations.

DR. HOWELL: Jeff's point's very well-made. This recommendation does involve an in-hospital screening. And again, our experience in that, in the only one we're doing, which is hearing, that mechanism has not been as crisp as we might like, as far as follow-up. So I think there are issues here.

Let me also point about implementation, is that many of the recommendations that we will come forth with in the future will have issues about implementation that can only be obtained when we implement them in large pilot studies. And then, because there are questions you cannot answer until you actually do it. And when we recommended that SCID be adopted, as you probably remember, we had almost the identical recommendations that followed -- if you look at the SCID recommendation and you look at this heart recommendation, they are almost identical at the end about who does what and so forth. And later today, we're going to hear about the SCID pilot project, which I think is just spectacular as far as the over 800,000 babies have been screened, et cetera, et cetera.

But anyway, I'm sure that there will be a lot of continuing interest in this area. And I think the key thing now is that -- and my final comments with Dr. Wakefield is that -- I asked her to be sure, from her job, to be sure that this committee met frequently and moved along as quickly as possible and that 90 days would be the outer limit and not the inner limit. And hopefully, that will proceed rapidly and we can look forward to moving ahead.

Are there further comments from the committee? I'm sure we'll hear some comments from the public when we get along.

We need to have Michele discuss the status of the annual report.

DR. LLOYD-PURYEAR: I wanted to give the committee a status on the voting and approval of the annual report. All members approved, except for the three new members, who weren't members during the voting process. So Doctors Lorey, Bailey and Thompson abstained. Dr. Dougherty and Boyle and Dr. van Dyck did not send forth a vote. But NIH and FDA did. And they approved. So there's a majority of committee members who have approved the annual report. Alissa Johnson --

DR. VAN DYCK: I had a concern.

DR. LLOYD-PURYEAR: Huh?

DR. VAN DYCK: I had a concern.

DR. LLOYD-PURYEAR: Yes, but Dr. van Dyck had a concern. And so, I wanted to --

DR. VAN DYCK: So the reason I didn't vote was because I e-mailed to Michele a concern I had about the annual report. And this is just to raise it as a potential issue. When I read the full report, I noticed that it had a part one and a part two. And the part two, where other elements of the law, other pieces of the law that were in addition to the responsibilities of the committee. And I was concerned, in reading it, that the committee has done so much work and so much good work and constitutes the bulk of the report in part one. And when I started reading part two, it was basically what agencies are doing, not what the committee was doing. And I had almost felt like it took away from the workings of the committee.

So the law says that we'll publish a report on peer reviewed newborn screening guidelines, including follow-up and treatment in the United States. And that part of the report, I think, is good and I have no problem with. My concern was only that it may dilute the work of the committee or seem to dilute the work of the committee by adding the work of the agencies to the report. So I did respond, I just didn't vote. But I responded with a concern. I just wanted to raise it with the committee.

DR. HOWELL: I'll make a brief comment about that, is that I think Peter makes a very good point in the fact that the material in the second part is not required for us to do by law. On the other hand, is that this committee is involved in a lot of activities and implementation. And again, I go back to SCID, where our work does move over into areas that are funded by CDC and NIH and pilot projects with the Newborn Screening Translation Research Network.

But I felt that those were really highly relevant to what we worked. And I asked Michele about this. And she did tell me that the agencies whose work was included there had approved it so that we were not doing something that was not approved by the NIH or by CDC. And as long as the agencies found the comments appropriate, I thought they were highly relevant to what we do, although they're not our core mission. This committee, one of the things I like about it is the fact that the committee likes to do anything that helps move newborn screening along at times when it might be a little bit out of our way. But some folks that read things religiously might not like the fact that we're a little bit out of our area sometime.

Coleen had a comment.

DR. BOYLE: I was just going to say, Peter, while I recognize that part one is responsive to the actual directive in the law, I might feel

similarly to Rod, that the work of the committee has really spurred on additional activities. And I think that was the intent of part, was really to show how the reach of the committee has influenced the field. So I personally felt that that was a nice complement to it. And the reason I hadn't responded yet is because we were still doing some changes to it, up to the last minute.

DR. HOWELL: Are there further comments about it? Well, then, I think you have the vote, Michele. Is that correct?

DR. LLOYD-PURYEAR: Unless, Coleen, do you want to --

DR. BOYLE: No, I'll vote to have it sent forward. I think we're comfortable with it now.

DR. HOWELL: And Denise?

DR. LLOYD-PURYEAR: Denise, do you have no objections to send it forward?

DR. DOUGHERTY: (No response.)

DR. VAN DYCK: And I am going to abstain.

DR. LLOYD-PURYEAR: So there's four abstentions.

DR. HOWELL: Okay, four abstentions.

DR. LLOYD-PURYEAR: Everybody else --

DR. HOWELL: And everybody else. So it goes forward, et cetera. I think that it's a report that's quite ambitious and it's showing what

all has happened, which I think is very good.

Anything else on that?

DR. DOUGHERTY: Alissa is going to go through the annual report just tomorrow afternoon, I think, tomorrow morning.

DR. JOHNSON: Afternoon.

DR. DOUGHERTY: Afternoon.

DR. HOWELL: We now move into -- we have a considerable amount of work on the public health framework for hospital-based newborn screening and committee discussion, which certainly is relevant to some of our other things. And first, the committee should review both the charter and the legislation before the discussion begin, and the briefing book. But let me make two points.

One of the questions is, does this committee have a responsibility to consider the cost of implementing our recommendations. And the affordable health care regulations that's located in your thumb drive points out that the comprehensive guidelines that are illustrated in the uniform panel of this committee went into effect at May 21, 2010. Plans and issuers are required, therefore, to provide coverage without cost sharing for these service for the first plan year in the individual market policy year that begins May 21, 2011.

So the point is that the plans are required, under the

Affordable Health Care Act, to cover the recommendation of this committee. And that's next week. Well, it's the 21st of May.

The subcommittee on follow-up and treatment is helping look at these relevant issues. And the first step is the following session that we're going to hear about. One is the role and responsibilities for screening, either the standard or care, of universal mandate and the role and responsibilities of public health agencies versus the general responsibilities of a public health framework. We may even need a reminder about government's traditional role and the public health as assessment, policy development and assurance. And since newborn screening is deemed an essential public health activity, then the assessment, policy development and assurance functions should apply. Albeit, they may vary among the various levels of government, local, state and federal.

And perhaps a couple of these questions are whether or not non-dried blood spot screening conducted in the newborn period should be deemed an essential public health service. Hearing is one, and obviously, the A.C. is the congenital disease being another -- or whether all government functions should be delegated to the private sector. IN other words, I think one of the questions that's come up is exactly what should be the role of the public health department in those hospital-based

screening programs. Where exactly does that stop?

The point of care testing has been defined as by the Follow-up and Treatment Subcommittee as medical testing at or near the site of patient care. As we end, I would ask the committee to review the legislation of the charter. And our charter has the following: "The committee is to provide advice to the Secretary about aspects of newborn and childhood screening and technical information for the development of policies and priorities that will enhance the ability of state and local health agencies to provide newborn and child screening, counseling and health care services for newborns and children."

And then, there is a long deal in the legislation that I will not read for you. But we are now going to move to the session that will directly address this. And we're going to first hear from Nancy Green from --

FEMALE SPEAKER: Oh, no, actually, I think --

FEMALE SPEAKER: We're going to say something.

DR. HOWELL: Oh, you're going -- okay. We'll hear from you two first.

FEMALE SPEAKER: We put the legislation in the book and on the thumb drive. And I think it was that legislation that was specific to the advisory committee was handed out this morning to --

FEMALE SPEAKER: It should be at your chair.

FEMALE SPEAKER: It should be at your chair. Just to remind everybody about the broad purview of this committee and some of the confusing aspects, perhaps, of the legislation of how you interpret it. So I'm hoping this discussion will help clarify some of those issues.

DR. HOWELL: Yes. And let me introduce Coleen and Jeff, who are going to be responsible for this session.

And it's all yours.

DR. BOTKIN: Thanks, Dr. Howell. And you've really done a nice job introducing the session for us. This really didn't arise out of the critical cyanotic congenital heart disease discussion with questions raised by a number of the stakeholders. So, you know, what's my role and responsibility with respect to screening of this type?

The committee has had a great amount of experience with newborn blood spot screening. And as recommendations move into these other arenas in which screening is conducted within the hospital environment, it raises appropriate questions about whose job it is to do various aspects of that and maintain the high quality of screening, universal screening for babies with these new modalities.

So I think the current questions that are being raised around the congenital heart disease statement simply reinforce the need to look at

this broader set of issues. So our intent is not really to focus on cyanotic congenital heart disease here, but rather the broader set of issues that are raised by screening that occurs beyond the traditional blood spot context, when screening is conducted within the hospital environment. Folks have raised questions, again, about, so what's my job, specifically related to health departments, but really, a variety of other stakeholders.

So our intent with this session is to hear from a variety of stakeholders about what the right questions are to be addressed in this context. So importantly, we're not going to try to answer these questions today that have been crafted, but rather to try to make a determination about what the right questions are, who the appropriate stakeholders are to have at the table as this conversation moves forward. And we presume this will be an active discussion over the next six, nine, twelve months or so as we try to adequately characterize the correct question to ask and how to answer those questions for this committee.

So Coleen has really taken a major leadership role with helping develop this session and conversation, as has Nancy Green. And certainly, our thanks to Jo Sugar as well for her excellent support for our activities here.

So what you see before you is a list of individuals who we've asked to speak quite briefly to this question of, are these the right

questions that we've drafted and who are the right stakeholders to have at the table as we move forward with this dialogue.

And the first presentation will be by Nancy Green, who is going to give us about 10 minutes of overview that will help us then hear from additional individuals who are going to provide perspectives from their professional backgrounds.

So, Nancy, thanks so much for everything you've done here.

DR. GREEN: Great. Thank you, Jeff and Dr. Howell and committee.

So, as Jeff mentioned, this is, sort of, the beginning. Or we've jumped into the middle. But this is not anything definitive. It really meant to, kind of, describe the landscape from a mile high. And maybe that perspective was inspired by the recent attendance of many of us at the pediatric academic meetings in Denver. So we'll stick to the mile-high theme.

And so that my comments are really a reflection of synthesis from the Follow-up and Treatment Subcommittee of this committee, as well as, I would say, the beginnings of input from some of the key stakeholders, but is not meant, by any means, to be comprehensive. Okay.

So with those caveats, okay, so I'm going to be talking about

the issues, the topics and the challenges around point of service screening. Actually, when I Googled the definition, it comes up as point of service testing. So we're going to have to, sort of, adjust our thinking about screening versus testing with respect to existing literature around point of service testing.

So the context of this session really is to address, you know, kind of, stepping back from these recent discussions, as have been outlaid in the comments by many so far this morning, the juxtaposition of really philosophical public health and pediatric concerns and then heightened by the recent committee reviews. And we've, again, begun to hear very clearly stated concerns by knowledgeable stakeholders about jurisdiction, mandates, financing and reimbursement. And I think all would share in the desire that roles, responsibilities, including, you know, fiscal responsibilities, are apportioned appropriately in the best -- in an ideal world.

Okay, so what is point of service screening? Again, this is taken from the referencing here. And, as Rod mentioned earlier, it's screening or testing at or near the site of patient care. So in this case, it would be the nursery. And the driving notion behind it is to bring the test conveniently and immediately to the patient. And I'll describe this a bit more later in the presentation.

And this point of service care or screening or testing increases the likelihood that the patient will receive the results in a timely manner. And I think, you know, as exemplified by the recent review of the CCHD, that the timeliness, I think, is the -- or urgency is one of the main aspects and that this is not true of dried blood spot screening. But while, of course, the blood spot is obtained in the nursery, that the actual testing or the screening is not performed there at the nursery.

Okay, so as the committee considers the interface between professional standards of care, public health programs, we are starting with newborn screening because, I think, we would all agree that these issues are broad and complex. And while we are cognizant that the committee has a broader role or broader charge in still thinking about childhood screening, which we just, sort of, agreed to stop the newborn because the issues become, sort of, logarhythmically complex very quickly. So at this point, the work group is fully focused on the newborn aspect of this screening, point of service screening.

And we all know that there are many professional guidelines in existence for screening of children within the context of child care, vision screening, lead screening, et cetera. And then the question is, how do these conditions differ. And, of course, we've discussed some of this before.

Okay, so I'm coining this term, "point of service screening-n," to remind us about the newborn or the nursery. But that being said, we really need to clarify roles and responsibilities and certainly, resources around this non-dried blood spot screening. And I think one of the key aspects to keep in mind is that those responsibilities and roles really may very much vary, depending upon the condition and the needs, both diagnostic needs and therapeutic intervention.

And, as, actually, Rod alluded to already this morning, that hearing screening is the only current example of point of service screening in the nursery. But I think there's broad agreement that it may not be the ideal example to follow, given the vicissitudes with uneven screening and reporting and follow-up.

There are certainly diverse opinions, depending upon where you sit and how your budget cuts look this year and also, I think moral issues about responsibility, for where these roles and responsibilities lie. Again, when we think about newborn screening defined as an essential public health activity -- and, again, Rod mentioned that public health programs may, in this case, be -- you know, we might want to think about, you know, should those public health programs be responsible for surveillance, evaluation and/or education, and leaving out, actually, a large part of the implementation.

Okay, so this is a grid that was donated by Denise.

So thank you.

We've adapted it a bit trying to, sort of, sort through, you know, what the factors are for these considerations. And so, if you just go across, the screening focus, certainly, you know, we're, again, talking about newborns, et cetera, the age of the child, the site of the screening, the site of the actual blood draw or hearing test or TCB or pulse ox or whatever it is, the site of analysis and also the site of follow-up and presumably, of diagnosis. And we haven't gotten too far into how quickly the diagnosis needs to be done, whether that even needs to be done prior to discharge.

The site of follow-up care may vary, whether it's, for example, in a traditional blood spot. Obviously, that's an ambulatory care, whereas in the CCHD, I think, the follow-up is most immediately in the hospital and then presumably, requires some ambulatory follow-up. And then the question about the role for public health programs with a question in the corner there.

So thank you, Denise.

So we also need to keep in mind that states vary in their response to those committee's recommendations once put forward and approved by the HHS Secretary. So, for example, we've heard from some

states such as in California.

Fred, correct me if this is not true. But that if the committee makes a recommendation, it's put forward by the Department of HHS. Then that becomes part of the mandate for California public health programs.

DR. LOREY: It's not in law at this time. But it was a law proposed.

DR. GREEN: Okay.

DR. LOREY: That's generally the way we try to do it.

DR. GREEN: Okay. You have to use your mike. Okay.

DR. LOREY: Sorry. That is not in law yet. There was a bill to that effect that was tabled. That's generally the way we, as the public health laboratory, proceed.

DR. GREEN: Okay, thank you. And we've also heard from Indiana that is tied to the law.

So there's a lot of clarity around definitions, I think, that that work has yet to be done. And, as I said earlier, that requires a number of stakeholders from several perspectives, including professional groups and the payers. And there's certainly no single right way or directive. As I said before, it depends on the condition and the state and then potentially additional factors as well, timing and urgency among them.

So we tried to then just get at our Denver perspective that bedside really would have to be justified on the basis of urgency, equity and/or efficiency, so, sort of, an additional push beyond the accepted definitions of the public health mandate for newborn screening itself. And so, I think the agreed upon definition for the point of service, again, would be prior to discharge of an infant, again, at least for the first screening test, not necessarily for the diagnosis. Although the assumption would be the diagnosis would be quick to follow or at least to diagnosis, to stratify those infants that would need emergent, maybe even pre-discharged treatment versus those that may be able to be followed-up subsequent to discharge.

And that the justification of testing and certainly, the justification about lack of parental permission would parallel those same justifications made for traditional dried blood spot newborn screening. So the issues, I think, then for point of service screening becomes both generic, in terms of the aspects of admission and specific for a specific condition under consideration in the context of public health framework.

And certainly, I guess, one could -- I think for some of the conditions, I think, you know, one would have to consider whether there's a public health role at all. Perhaps for some conditions, there might not be a public health role. And, you know, what would the minimum public health department role be? And that may be, again, at a minimum,

centralized data reporting and program evaluation. And, you know, whether that would fit with any of the conditions that have previously been considered or to be considered, I think, you know, remains to be seen.

So we decided to, again, step back and think about the key attributes. What would discern point of service screening versus something else? And, of course, the overriding issue would be that immediate diagnosis and follow-up care are likely to be needed and provided so that the condition -- so, again, thinking about, sort of, the three categories of newborn screening consideration, the condition, the screening and diagnostic tests and the treatment, for the condition.

The diagnosis would have to be a serious condition. And I think Ned has drilled in my head, anyway, the issue about serious, what that means. And that needs to be made prior to discharge and potentially prior to discharge before initiating treatment. The diagnosis must be interpretable within the newborn period. And, as I said before, at least to stratify categories of risk. It may be that some infants would screen positive, and yet be found to be of low risk and, therefore, subsequent evaluation and treatment could wait.

So thinking about attributes that would be specific potentially to point of service screening for the test, they're not so different from traditional newborn screening. But I think it's worth going through them as

we think about non-dried blood spot modalities of screening. So clearly, the screening tests have to be easy, easy for the -- you know, not taxing for the infant, you know, don't require a general anesthesia for a general MRI or something like that.

They have to be reasonable, safe and acceptable to the community, and community to be defined. These procedures have to be relatively simple and quick for the staff. So, you know, that's a manpower issue and instrumentation issue for the nurseries themselves. The results, I think without exception, the results of the screening need to be promptly obtainable on-site.

It doesn't necessarily mean that the testing is -- you know, the screening assay is done on-site, particularly for potentially, like, a small nursery or a rural nursery. But I think that would be the goal, would be to have that screening performed and interpreted on-site -- that the screening test would be available with reasonable investments of nurseries, again, of diverse sizes and configurations and that there ought to be some standardization, both locally and more broadly, for that screening and that quality assurance is available locally.

So as far as, then, the subsequent diagnostic tests or procedures for these kinds of screenable conditions, that -- this is actually the availability of the diagnosis came up, I think, with the CCCHD and the

issue of a pediatric cardiologist, echo-stenographer, which is an issue that one would hope that the -- one would plan that the diagnostic test or process for testing would be available either at the site or transportable.

That is up for more discussion -- that the diagnostic testing would be feasible and definitive, at least, again, for those who are at most -- infants who are at most imminent risk of harm from the condition and that the diagnostic testing would be safe, safe for those with a false/positive screening and relative to potential benefits -- or in proportion to the potential benefits for those who are true positives and with a favorable ratio of potential benefit to cost.

Okay, so when we think about, going back to the previous discussion from this morning, the current point of service screening and, you know, we'll have to see what happens with CCCHD. It sounds from the comments like the recommendation of the committee will be taken seriously and will need some more around implementation. But I'm not going to address that.

And again, I think the underlying assumption here is that -- has to be -- that state health departments may bear less responsibility if the point of service condition is added to the recommended panel than they currently do for non-dried blood spot screening. And then, I think you've heard some of this today.

So what are the considerations for this committee for point of service screening? And, you know, could the current criteria that this committee uses or additional criteria and structure for review be used to distinguish tiers of recommendations, each requiring different levels of public health involvement, as I mentioned previously? So what entities would be responsible for follow-up treatment and tracking? Could those roles be distributed elsewhere? And again, to emphasize, could public health programs take on more limited roles for these non-dried blood spot screenings?

So just some thoughts, then, to wrap up. And the first was donated by Chris Kus.

So thank you for those comments -- that the criteria used by this advisory committee for universal screening may differ from other and potentially, although not necessarily, more stringent criteria, from those used in public practice, for example, the U.S. preventive task force and others; that there needs to be an interface between professional standards and public health programs. I think we all would recognize that -- and that the advisory committee should certainly seek input from relevant professional organizations, service providers and hospitals, among them. And then what gaps in the funding streams need to be addressed if universal point of service screening for newborns became standard of

care?

So, as I mentioned, this is some early thoughts from the subcommittee. This represents an opportunity to begin to think about that and more collaborative thought is needed around the issues that we have outlined.

And, you know, one of the options for the committee to consider is whether it would be useful in moving forward whether a meeting of key stakeholders should be convened to begin to grapple with some of these issues and get a little lower than a mile high or higher, however you look at it. And then, Coleen had pointed out there may be some opportunities for publication on public health professional journals as well as we consider how to move forward and how to disseminate the thoughts of the committee.

So that's it. Thank you for your attention.

DR. BOTKIN: Thank you, Nancy.

I think what we'll do is accept just a couple of questions at this point.

DR. BAILEY: So thanks for that very helpful overview. There's one question that, I don't know, maybe it's my misunderstanding things. But on slide 11, you say that results promptly obtained on-site. And I think you said -- I thought I heard you say that that was not a

necessary criteria, but that was something that would be an aspiration. But to me, that's what the fundamental essence of point of care screening is. Otherwise, it's not point of care screening. Am I misunderstanding something?

DR. GREEN: No, I don't think, Don, that you're misunderstanding at all. I think we would agree with you that it -- but I'm -- since the landscape is, sort of, before us and lacks definition, I hate to say always. But it's only at that caveat. But I think your comment reflects, really, the perspective of the group. Thank you.

DR. BAILEY: Thank you.

DR. BOTKIN: Alan?

DR. FLEISCHMAN: Nancy, thank you. I think it's really an excellent beginning. But I would like to caution us to separate pragmatism from principles. And you've, kind of, interdigitated them a great deal here. And I think if we can hold to principle at the outset and try to figure out from the children's perspective what's the right thing to do, based in the principles of newborn screening, which were used in the creation of the original panel, which were used in this committee's deliberations and once we make a decision what the right thing to do is, then we have all kinds of pragmatic implementation issues that we need to deal with.

And I think we have to hold whatever system accountable.

And certainly, the clinical practice system of standard of care is not accountable in the same way that mandatory testing of every individual is accountable at the public health perspective. So I understand all the pragmatic problems. And I'm very sympathetic to them. But unless we start in principle, I think we will get really embedded in the pragmatism at a level that won't allow for constructive and rapid resolution of the issues.

DR. GREEN: Oh, I think your points are very well-taken, Alan. And I appreciate them. And, you know, I think, on the one hand, you know, figuring out which is the cart and which is the horse is very important. And, as we've heard earlier about SCID, that, to some extent, the committee's decision really becomes the horse. On the other hand, I think we're all cognizant of the issues around, for example, hearing screening, which is not life-threatening and, you know, maybe not a good way to hold up the concerns about pragmatism. But I think your points are important. Thank you.

DR. BOTKIN: And we will have additional time at the end here for open discussions. We want to go to our list of stakeholders here. And again, the question that's been posed to these stakeholders is, what are the right questions to be addressed by this larger debate, and do we have all of the correct stakeholders identified to participate in that discussion as we go forward with this.

So Dr. Howell's going to be our first, unless you feel you've already addressed this set of issues with your previous comments.

DR. HOWELL: I think I've already addressed it. I think that the bottom line is that if you look at the legislation, which you all have at your desks, the thing that's, I think, the key thing is that we're supposed to provide such recommendations, advice or information as may be necessary to enhance, expand or improve the ability of the Secretary to reduce mortality and morbidity from heritable disorders, which may include recommendations, advice and information. So it's a very broad charge, basically, of things to be done in the newborn period that can reduce mortality and morbidity.

DR. BOTKIN: Okay.

Any special questions for Dr. Howell at this point? All right.

Then, from a primary care perspective, we have two individuals who have been willing to help us out with this conversation. And we are looking here at a series of questions that have been drafted. So hopefully, all the committee members have an opportunity to look at those. And thanks to the subcommittee and also to Michele and Rod for helping refine these. And we want to refine these further.

So our first stakeholders are primary care providers. So Fred Chen from the AAFP.

Fred, thanks.

DR. CHEN: Thank you. And again, we're happy to represent the American Academy of Family Physicians. Family physicians and other primary care providers are an important part of the newborn screening process, follow-up, diagnosis, treatment. And so, I think that's why we've been represented at this table and appreciate this opportunity.

You know, the reality of primary care practice, both family medicine and other specialties, is that we have a lot of clinical practice guidelines to deal with. And then, they come from various specialty societies. They come from government sources. And then, there are the ones that are actually mandated by law. And newborn screening is, of course, the example there.

The majority of clinical practice guidelines around prevention and screening, at least for family medicine, are propagated by the U.S. Preventive Services Task Force. And the American Academy has had a long history of working with that task force and being supportive of their evidence process and, in fact, then, taking each of their recommendations to the academy itself and evaluating them and then recommending them to our members. So there's only, sort of, three messages that I -- three comments that I have about this.

One is that all of our clinical practice guidelines -- there's a

policy at the academy in terms of looking at clinical practice guidelines that they oftentimes -- that they always need to be balanced by individual clinical decision making and the ability of physicians to make clinical decisions in individual contexts. And so, that may be an important distinction between mandatory newborn screening and what we think of in a broader sense as clinical practice guidelines.

The second is what this committee's already done, which is embrace evidence-based methodology and decision making around recommendations. And so, we are strongly supportive of that process. I would say that this formulation of screening in terms of point of service or point of care screening is a little bit odd in that, in my experience, very few of these guidelines are location-dependent. And so, it does raise some questions about, sort of, the whole framework of how we think about this particular question and whether or not point of service or point of care is really the right thing.

And the last thing I would say is that one of the ways that the U.S. Preventive Services Task Force has been so successful in its work, in addition to having a strong evidence-based methodology, is that it has been very clear about who its audience is. And its audience has always been practicing primary care providers who are delivering preventive services of some kind or another. It does not make recommendations to

states. It does not make recommendations to public health laboratories. And so, I would think that, in a way, that's been part of its success.

CDC has similar success in its clinical preventive community guide. And so, that might be a -- I think that's also an important thing for this committee to think about, is -- and that's how we got into trouble, is, sort of, the precision around the audience and who we're making recommendations to.

DR. BOTKIN: Thank you.

Quick questions, clarification for Dr. Chen?

Okay, Tim Geleske from the AAP?

Thanks, Tim.

DR. GELESKE: Thank you, Jeff.

Well, I would certainly agree with everything that Freddie said. I think there's multiple factors that influence what a primary care physician does in their practice, including political recommendations, cultural things, family factors. But probably most importantly, we followed the recommendations of our professional societies.

In pediatrics, Bright Futures, which is the guidelines for health maintenance in primary care pediatrics, actually recommends nine different universal screens as part of our exam, which includes dried blood spot and hearing screening. Some of those have stronger evidence bases

than others. And probably the most common thing that we do, the physical exam, has very little evidence base, but yet, we all carry that forward. And I don't think there's anyone here who would disagree with doing a physical exam at a checkup.

But we really depend on our leadership. Most of us, I don't think, have the resources to evaluate the data and make evidence-based decisions at our practices. And we really look to our leadership, I think, particularly in the academy for guidance. We look to them to put together the experts who know that literature and the evidence base. Our clinical guidelines, I think, oftentimes have a lot of practical experience worked into them. A lot of expert opinion is also there because there's just not the evidence to support everything we do in primary care.

But the lack of evidence doesn't mean that there's a lack of usefulness. And it's still very important, the things that we do on a daily basis.

I think, in terms of what would mandate a universal screening from the standpoint of this, is the things that have guided us for all of our decisions. You know? It's an accurate, valid test to do it, that the benefits, by far, outweigh the risks of doing the testing and that there's some treatment modalities. I think urgency is important. So for a point of care testing, to have that result right there, as you mentioned, I mean,

that's the whole idea.

And so, you know, I guess, as Alan put it, the principles are very important. If there's one thing that's come out of the pulse oximetry and critical congenital heart disease discussions, is that the agenda for that has really been pushed for a very critical need. And this will save lives, whether it's adopted by the Secretary or not. But now it's out there. And the AAP is endorsing it. And it's going to be published. And so, some of these clinical practices will start to be put into effect. So I think that's my comments.

DR. BOTKIN: Great. Thanks, Tim.

Any specific questions for Tim?

Alan?

DR. FLEISCHMAN: Yeah, I just wanted to ask both Fred and Tim a question. We have this kind of magic idea about what standard of care is. And I just wondered, if we took all the pediatricians in America -- and that would be a good thing, that we look at all them -- and all the family physicians and we reviewed their compliance with all of the standard of care measures that you've just outlined, what percent do you think we'd have 100 ascertainment of such measures? And maybe it's a rhetorical question.

And the other question is, what is the standard of

accountability in the clinical frame. Lawsuits after deaths? You know, so I think we have to -- when we're comparing accountabilities, I think we really do have to understand the very important nature of the clinical practice guidelines, the Preventive Services Task Force recommendations and the rarity of the disorders we're talking about and the need for 100 percent ascertainment of the screening tests.

MALE SPEAKER: I mean, Elizabeth McClen did a study several years ago looking at delivery of preventive services to adults. And it was 50 percent, 50 percent of American -- but, having said that, I mean, there is a lot of room for individual practice variation. And what we're really talking about is that -- and the crux of your question is, sort of, when is something mandated by law and when does something fall into clinical practice.

DR. GELESKE: Yeah, I would like to say that ours are 50 percent. I doubt that. You know, the Bright Futures guidelines is about this thick. So I'm sure that most of us don't do everything that's recommended by our academy. But it's the critical nature, you know, the urgency of the condition, I think, which should drive the decision for mandated universal screening.

DR. BOTKIN: So these would be characterized as aspirational standards, then?

DR. GELESKE: I think that's not inappropriate, yeah. I think someone has to drive the discussion. And I think we've driven the discussion on congenital heart disease, not that other people weren't looking at it. And certainly, advocacy groups were certainly pushing it. But the agenda has moved forward rapidly in the past year since it's come up.

DR. HOWELL: Jeff, I think it's important to point out is that most of the things that we're talking about are not mandated by law. Most of them are recommended and so forth. There are a few laws, but most of it is very strong recommendations.

DR. BOTKIN: Good. And I think, clearly, part of that, too, is the existence of accountability structures. You know, as Alan talked about before, blood spot screening folks are collecting data about.

DR. HOWELL: Absolutely.

DR. BOTKIN: Whereas in these other contexts, the recommendations are out there, but nobody's actually held specifically to those standards.

DR. KUS: Can I just make a comment about that?

DR. BOTKIN: Chris, yeah?

DR. KUS: Because I think there is evidence -- there is work about that, the core measures that are being reported that Medicaid

programs and state child health programs have performance measures. One of them that's proposed a developmental screening. One of them is a Chlamydia screening. And states do report on that. So in our state, we have percents of it. But none of them are close to 100 percent. And part of the reason is that, by reporting on that, we're stimulating people to where do we do improvement. So public health has a role, even in the clinical part.

DR. BOTKIN: Good. Well, and it seems that part of our question here, of course, is is there something that this committee's recommendation implies a translation into a different sort of accountability system. Do we intend to say that, by virtue of being on this panel, somebody will be collecting data about nurseries and clinicians that may be different than, say, professional standards, Bright Futures, where there's not that sort of accountability system.

Okay, yes?

DR. KUS: One follow-up comment. You know, I think it's important to recognizes in all of these discussions that what we're really talking about is a point in time. And one of the things that we haven't done very much with this committee and in terms of looking at the disorders on our panel is the evolving evidence as screening programs move forward and whether or not we should be making a decision to actually remove

something from the panel. So I'm not advocating that we do that for anything that's out there now. But we do have to recognize that, although expert opinion and experience are not the best evidence, they are evidence. And we do the best we can with the evidence that we have.

But, you know, 200 years ago, bloodletting was standard of care and based on experience and expert opinion. We've evolved past that because we reevaluate the evidence as it comes forward. So, you know, if we make a recommendation and, in spite of our best efforts, we get it wrong, we have the opportunity to go back and readdress that. And so, that may be even more important as we go forward with some of these point of care screenings because we have so little groundwork that we can fall back on and experience to help us.

DR. BOTKIN: Yes?

DR. GREEN: Chris, I wonder if you could just maybe elaborate a little bit more because I think a key point that people keep coming back to is accountability. And there's a very big difference from mandating something and recommending it. But I think you were making a point a moment ago that there is data collection for things that are not mandated. And I think that's a key point. And I wonder if you might elaborate a tiny bit.

DR. KUS: I could do it in -- I'm scheduled a conference

(inaudible).

DR. BOTKIN: Okay. Excellent. Thank you.

All right, Joe from the Hospital and Specialty Care perspective? Thank you.

DR. BOCCHINI: Okay, thanks. First, I want to thank Nancy for a very nice presentation that really framed this discussion quite well. I think the questions that are being asked are very appropriate. And I think that I wouldn't separate the universally-recommended newborn screening test from good clinical care because I think it's the same thing. I think there's a different evidence base perhaps and a different level of support for that recommendation. But I think they're the same. And I think that the roles and the responsibilities of individuals really depend on what is being looked at and what the potential outcome is for that.

And I think if we take the critical congenital heart disease as an example, I think if approximately five children are coming to the emergency room each day in extremis because of a potentially diagnosable congenital heart disease in the nursery, that's a public health issue, to me. That's something that a mandate or a recommendation that comes from the state that then requires nurseries and physicians to follow a set of guidelines, I think, can solve this problem more rapidly and more effectively than using guidelines and that come from different stakeholder

groups.

However, I think that in developing those guidelines, it's critical to have those stakeholder groups involved. So I think one of the key issues, as we look at point of care testing, is what stakeholders need to be involved. Hospitals obviously need to be involved. Public health people need to be involved, and the different physician and other stakeholder groups as well, to formulate an appropriate set of implementation guidelines to make things work. But I like Dr. Fleischman's comment that the principles are important. And then, you can't figure out all the implementation issues in the beginning. And once we figure out what needs to be done for children, then the implementation can follow and the roles and responsibilities can be divided.

We have a number of other public/private or public health and physician-related guidelines that we do together. We do lead screening. That's a public health issue, but it's done in individual offices. And it's a requirement for early periodic screening and testing at the EPST program. So I think that there are many examples that we could use that could be used to, sort of, develop what we need to make this work. So I think public health can have a significant role in point of care testing. Again, depending on the issues, it may vary. The role may considerably vary.

Clearly, the hospitals are different here as opposed to a blood test on a card, which is then sent to the state to have testing done. Here, the hospital has to have people who are trained who can do the test effectively. They have to have the equipment. And they have to have the ability to act on the results in an appropriate way. So I think the responsibility is very different.

However, the public health people can put the infrastructure together to educate, to provide informational things to parents and to physicians and then, give, perhaps, smaller hospitals, that don't deal with this on a regular basis, the contacts they need, if there is an abnormal test, to get to the right pediatric program, wherever -- for example, if it's congenital heart disease we're talking about. So I think that there's a considerable role. And I think that the questions are being framed appropriately.

DR. BOTKIN: Great. Thank you.

Questions?

All right, let's turn to public health perspective. And Chris is going to lead off for us.

DR. KUS: Sure. Chris Kus, and I'm representing the Association for Stated Territorial Health Officials. So that's the health departments. And it's important to know that newborn screening programs

are within the health departments.

And I would start out by saying that the whole concept of point of service or point of care is a red herring because I don't think that's the issue. I think the issue is what we're talking about, is universal newborn screening and the other types of screening are clinical screening. When you look at Bright Futures, which is the guidelines for pediatrics, they talk about universal screens, and they talk about targeted screens. But I think Rod made the point. When we're talking about something being a core condition, it then leads often to state-mandated programs. And state-mandated programs inherently says public health is involved in that.

And I think our questions reflect the difference of terminology. Because if I look at the public health questions, the first question says, what do public health departments view as the responsibilities in state-mandated newborn screening. Well, ASTHO has a policy statement out about that that's classic public health. That's what we do: assurance, policy development.

And then the second question says, what are the responsibilities of the state public health department in point of care hospital-based screening. I don't understand that question because I think the first question is what the issue that we're talking about. And to talk

about -- is that when we're talking about state-mandated programs, health departments are responsible for that population-based data that then feeds to the national data that leads to the evaluation of the program. And I would highlight for folks one of the difficulties when we confuse that.

People mentioned hearing screening program. And I think part of the problem with the hearing screening program is that it is mixed with early intervention program. And a lot of times, it's not looked in a public health point of view. And therefore, that's why our numbers are lower. So that's one part of it.

Then I do think, with regard to clinical standards and clinical screening, there is that movement for accountability about that. We're talking about electronic reporting. We're talking about reporting on outcome measures like Chlamydia. And state health departments, at least in the New York State Health Department, where Medicaid and the state child health insurance program are, we put out reports about how plans do relative to those screening, which are certainly not 100 percent and there are different levels. And we then use that to talk about what quality improvements that you need to do.

So, you know, my main point would be we're confusing the things like putting in the concept of point of service testing because it's really universal newborn screening testing versus clinical testing or

screening.

DR. BOTKIN: Thank you.

Questions?

Okay, Fred Lorey? Thanks, Fred.

DR. LOREY: Well, I guess, I thought we would be of the same mind. But I guess I'm going to have to disagree with Chris.

DR. KUS: But that's just a part of the health department, just so you know.

DR. LOREY: That's exactly my point. I think we have some issues with definitions here, as I've been listening today. On the public health, from the public health point of view, I think there's no question that public health needs to be involved in this. There are various parts of public health, one of which is newborn screening, which is laboratory -- public health laboratory-based screening on blood spots for heritable disorders. And heritable is a word that's in these guidelines as well.

Newborn screening has traditionally been defined in this way. The way the panel's recommendations came about, until heart disease screening, have been interpreted that way by people involved in newborn screening. So I think there is a difference between newborn screening, as it has been defined, and other screening, that I do not consider part of newborn screening, unless we change the definition.

Now, one of Nancy's slides -- that was a great overview. But I noticed a couple places there, one where she was defining hospital-based procedures. And it said diagnostic testing. And then beside that, she had put in parens, screening. Well, they're not the same thing. You know? We're not doing diagnostic testing and newborn screening.

The one thing I actually wanted to start with was I have no issues whatsoever with the review process, the scientific review, the recommendation that screening, if we just use that word, is appropriate for congenital heart disease. But, quite frankly, I don't think it's appropriate for public health laboratories. I don't think it's in our purview. It's not in our state statutes.

And I've heard also that, you know, we can't separate -- Alan said we can't separate practical from the scientific assay. But you have to. As a person from the practical end, we're the ones that have to implement this. And you have to consider that. Because if you make a recommendation, and everybody's been looking at newborn screening, in the committee, in the narrow sense, then all of a sudden, our charge has changed.

And, quite frankly, we don't have the money or the staff to expand what we're doing from what we're doing now. We don't have it. We take a lot of pride in quality control of our assays and our data. We

don't take other people's data that we have no control over and follow on that.

Just to give you an idea of what I've done -- what I've spent a lot of my time on the last couple of weeks and why I have less hair than before and greyer than before, both related to the committee's work -- maybe this will put it into perspective for you. SCID screening -- perfect example of something that should be in newborn screening, just a perfect example, great treatment, great test, no question that it should have been recommended.

So what I've been doing in California -- although we've had a very successful pilot that we were only able to do because we didn't tell the folks we were supposed to tell and we were fortunate enough to get, you know, some financial assistance from outside the department. Now there's legislation proposed to make it permanent. And when I got off the plane last night, I saw a report from the Budget Committee on the funding.

Even though they passed the bill that didn't contain funding issues unanimously -- they say, "Oh, yeah, it's a great idea." We have to deal with the budgetary issues. And the e-mail said their analysis of it was it would cost about $7 million, which is probably about right, but that the savings would be about $1 million dollars a year. Well, I have Dr. Buckley sitting here next to me. And I'm sure she'd probably be the first person to

say, "Well, yeah, maybe you'd save that much on one kid, but not 12."

So we're having to deal with people who make monetary decisions who, you know, don't really care what we have to day about the disorders, even, to be frank, what the committee says, because, you know, they're not looking at that. They're looking at money.

Now, the other thing I've been dealing with is a media request from CBS News regarding a story they're working on with Hunter's Hope Foundation on Krabbe Disease. And the reason we were approached is because there was going to be a family with Krabbe on this story. And they keep asking us things like, well, why does California not screen for Krabbe. And we keep saying, it's not recommended by the committee. The question is, why is New York the only state screening for Krabbe.

So whether it's really perfect recommendations that the committee does or decisions that they make -- and I agree with the decision that Krabbe is not ready, when you get down in the trenches where we are, we have to deal with other things. And if the role of this committee is expanded to hospital-based tests, which is very different from what we've been doing, we're going to be in a position of, a, not having enough money, but having to make choices. You know, we can only afford to add one disorder at a time.

So, you know, whether it's Krabbe and having to, you know, be sympathetic to those families about why we can't do that, but we can do SCID, or the heart disease people versus Pompe, which may be recommended by the committee, you know, we can't do it all. So I agree it's in the public health venue. I don't think it's a newborn screening public health laboratory venue. That's my personal opinion.

In California, newborn hearing screening is not even, not only not a newborn screening, it's not even in public health department. It's in the Department of Health Care Services.

So I guess what I feel is if the committee is going to go down this road, you have to include the practical things from the beginning, or the committee has to include in its recommendation how the implementation should occur. Because I agree with what Dr. Van Dyck said earlier in terms of the Secretary's interpretation. You know, it's nothing to do with the quality of the recommendation and the work that was done. It's the implementation. But you can't ignore the implementation.

DR. BOTKIN: Great, thank you.

DR. LOREY: I've probably got lots of enemies now.

DR. BOTKIN: A couple of quick questions?

DR. LOREY: But I will say that every newborn screening

person that's in the trenches, like me, I've spoken to is of the same mind as me. So, you know, you need to listen to us.

DR. BOTKIN: Jane, did you have a question?

DR. GETCHELL: Well, yeah, kind of a follow-up to what Fred was saying. I'm beginning to understand why this would be in the public health purview. But I think in coming to that decision, it's very important that we determine what exactly we mean by that, what would the role of public health be. Because I think one of the things where a laboratory could come into it is assuring the quality and the performance of this particular test. Is that going to be part of the public health role or not?

And the other thing I wanted to mention, yes, we have many diseases that are reportable to public health, Chlamydia being one that was mentioned as well as newborn screening. But what we do with those results is very different. It's far more time-sensitive, newborn screening follow-up, than, for example, Chlamydia follow-up. Just something to be aware of.

MALE SPEAKER: Could I just make a quick comment about that? Because the comment is we get Chlamydia reporting, but we also get information about how well Chlamydia is being screened for, which is the clinical part.

DR. BOTKIN: Jerry?

DR. VOCKLEY: Thanks, Jeff.

A couple of comments -- first of all, I do have to disagree with you, Fred, that this committee is not constituted to look at blood-based, laboratory-based newborn screening and newborn screening tests. There's nothing in the law that limits it to that. And I don't think it's appropriate to be limited to that.

I do agree we have to consider implementation. And when we do the evidence-based review and make our decisions, there are explicit questions in the decision matrix that focuses on the availability of testing and, as best we can divine it from the literature and experience, the kinds of costs that are involved. However, we're making recommendations to HRSA. It is impossible to take into consideration the specific implementation challenges that -- and practices -- that 50 states are now going to have to use to decide how they're going to implement that.

Part of what we do -- part of what we have done has been to push the limits. And pushing those limits, in some sense, also requires that we go into a zone that may not be comfortable at any one point in time for some of the interested stakeholders. But these can be used, these guidelines can be used as leverage to the other state legislatures to

lobby for additional funding. It may not be reality right now, but eventually, that's the kind of impetus that these kinds of guidelines would bring.

DR. HOWELL: Can I make two very quick comments? I know time is a problem. Fred's points, I think, are very well-made. I think that there's absolutely no question that it's in the purview of this committee to look at newborn screening, other than the dried blood spot. I think that the responsibility of the state lab, in other words, Fred's group, is where the issue is, you know, how will they deal with this and so forth. And it may not be a responsibility you personally are dealing with. I think that's the key.

The second thing is is that we've never made a recommendation that had more detail recommendations about implementation than occurred with the congenital heart disease thing. And again, many of you have seen the paper.

You have not probably, Fred, but that has a very detailed plan for implementing that and so forth. So I think your point's well-made.

DR. LOREY: Yeah, I actually don't disagree with either of you about most of that. If that is in the purview and that's interpreted as the charge, we need to change the definition of newborn screening because that is not the traditional definition of newborn screening. It's not how it's been perceived by the public. And we do have to deal with the

legislatives. And if they're giving us a hard time about screening for SCID, what do you think they're going to do about these hospital-based tests? That's just the reality for us.

DR. BOTKIN: Okay, let's pick up on this larger conversation. If we end up having some time at the end -- we still have two more speakers with us, I hope.

Ned, are you on the phone with us?

FEMALE SPEAKER: He actually -- and I don't know why you weren't communicating with -- he can only join tomorrow morning.

DR. BOTKIN: Oh.

FEMALE SPEAKER: He had a personal emergency.

DR. BOTKIN: Okay. Well, we certainly will welcome his participation in future discussions here.

Yeah, Ned, of course, has been a longstanding member of this committee, but also has chaired the U.S. Preventive Services Task Force. And so, I think the set of questions that are part of that discussion have to do with other evidence-based review bodies that are out there that are making similar sorts of recommendations. So I think part of the larger discussion will have to be where does this Secretary's Advisory Committee fit with respect to other advisory groups out there related to screening and also, how is it that recipients of these recommendations in

the clinical environment view these sorts of recommendations.

So, you know, as we've seen with some of the background information, the U.S. Preventive Services Task Force has a fair amount of respected leverage with respect to what happens in the clinical environment. So I think we want to think about this committee's role with respect to other similar bodies that are out there.

James Figge, I think, also was going to be on the phone with us.

James, are you with us?

DR. FIGGE: Yes, I am. Can everybody hear me?

DR. BOTKIN: Yes, thank you. Go ahead with your comments.

DR. FIGGE: Good morning. This is Dr. James Figge. I'm a medical director with the New York State Department of Health and the Medicaid Program. I also sit on the Follow-up Committee of your committee. And I wanted to make some comments today pertaining to the role of state Medicaid agencies in your deliberations.

Medicaid agencies, as I see it, have three fundamental roles in their various states. The first would be their primary purpose, which is to provide insurance coverage. The second is that they have a public health role. And they can use their marketplace power, which can be

quite considerable in some states with policy and coverage development, to implement various key public health objectives on the ground. And third, Medicaid agencies have a regulatory role.

I see Medicaid agencies, ideally, placed at the intersection between the traditional public health functions such as newborn screening and the traditional medical delivery system because Medicaid can traverse back and forth between both systems and take some of the output from public health and put it into practice in the general medical practice system. Let me give you an example of how New York Medicaid has done that.

We've worked very closely with our newborn screening program. And, as you may know, in New York, Medicaid is part of the Department of Health, although a different office than the Office of Public Health. But we have worked very closely with our newborn screening program to ensure that children who have a positive newborn screen will be able to move on to the next step in terms of getting the appropriate confirmatory tests, if they're in the Medicaid program.

So we have worked very closely to develop coverage guidelines for Medicaid, which anticipate the needs of those children with the positive newborn screening to ensure that they can go onto the next step and get continuity of care, get the necessary follow-up testing that

they need. So that's what I'm talking about in terms of the intersection between the traditional public health and the medical delivery system.

So we're able to span both of those domains and to provide continuity. And so, I think, for that reason, Medicaid needs to be at the table for these discussions. Not to mention, of course, that the patient population served by Medicaid in most states is very rich in newborn and older children. For example, in New York State, we cover nearly half of all the births in the state, you know, which is a tremendous impact on, you know, the operationalization of what you're considering here. Because basically, if Medicaid implements something on the ground, it's going to be in place for nearly half of the newborns in the state right up front.

And some of the comments that I'll make now may also pertain to private insurance as well. But I wanted to point out the unique positioning of Medicaid in this discussion.

Now, in terms of Medicaid and insurance function in general, I think it's very important that the committee work with the new ACA framework. And it's very important that whatever you develop, you know, be incorporated into the comprehensive guidelines that will be supported by HRSA so that it gets into the ACA framework. That will be critical for Medicaid agencies and other insurers as well.

And I know some of the previous speakers have talked

about evidence-based medicine. That is also very important for Medicaid. Many states are involved in collaborative where there is extensive evidence-based work done on a coverage policy for Medicaid programs. Almost every Medicaid agency that I know of has medical directors that are working very hard on making sure that their coverage policies are evidence-based. And so, we place very high priority on evidence-based work that comes from various HHS agencies, including, but not limited to, HRSA, CMS, CDC, ART, FDA. And in New York, we have worked very closely with a number of those agencies on some of our own coverage policies.

And other speakers have talked about federal panels such as the U.S. Preventive Services Task Force. I can tell you that the work done by that body is viewed almost universally by Medicaid agencies and most other insurers as well as being, you know, an extremely excellent example of rigorous methodology. The task force uses very good methodology for classifying the strength of evidence. They provide a very transparent summary of the evidence in great detail, which gets published in peer-reviewed literature, traditionally, and high-impact journals such as The Annals of Internal Medicine. And their work is very highly regarded throughout the insurance industry.

So I would recommend to this group that you consider using

that model of evidence development, transparency, publication to really get your work out into the public domain. That will help with the adoption by Medicaid agencies and other insurers. And those groups, taken together, can really help with the implementation. And even Medicaid can help with the enforcement, since we do have, in each state, regulatory authority as well.

So I think, you know, Medicaid agencies can be a very, very major asset when you consider rolling out some of your new guidelines. So, again, I would recommend that the committee adopt the type of model that the U.S. Preventive Services Task Force uses for the evidence development, the transparency, the publication. And I think Medicaid agencies, you know, would use that as a very good standard when they are developing their own coverage policies. And other insurers would as well.

And I wanted to make one final point, since I also sit on the Office of the National Coordinator HIT Policy Meaningful Use Work Group. We have brought forth to an HIT Policy Committee some objectives from your group. And there is definite opportunity for a cross-pollenization between what you're working on and what OMC is working on in terms of developing meaningful use standards for electronic health records. And that's another place where you can develop electronic standards which will

help get some of these things done as there is greater and greater adoption of electronic health records and point of care decision support. Those types of tools can help clinicians know what the evidence is and also understand that there may be an obligation to get some of these things done. So I just wanted to point that out as an opportunity for collaboration with yet another agency under the HHS umbrella.

DR. BOTKIN: Great. Dr. Figge, thanks very much. Let me see if there are any specific questions for you.

DR. HOWELL: I'd like to make a comment. And that is, as the committee is aware, the evidence group working with this committee follows a highly-rigorous evidence pattern, as you know, that's organized under the aegis of our evidence review group at Harvard. And again, the pattern of evidence review has been published in peer-review journals. And the evidence reviews that come to this committee have also always been published in high-impact journals so that we take that recommendation seriously. And I think the committee has followed that, clearly.

DR. FIGGE: Yes. And I'm not doubting that. I just wanted to state that, for the record, that continuing that pattern, you know, will have, you know, significant impact on the insurance industry. And I just wanted to make sure we stated that, for the record, and didn't take it for

granted. I'm very well-aware of the work that has been done by this committee, so I'm not doubting that for a second.

DR. BOTKIN: Mike?

DR. WATSON: Yeah, only to expand on something I said at the last meeting, and I was asked to engage discussions with joint commission that were extensions of some discussions we had had under some previous activities. And, you know, they bring some fiscal accountability to the table for hospitals. And, you know, they've become very interested in newborn screening, in general. They recognize it is fitting into one of their major mandates of a critical result value that, you know, there's an obligation to act on, which can tighten up one of the loops in newborn screening and perhaps have an impact on the hospital-based screening activities.

Right now, the last I heard from them was about four weeks ago, where they were trying to balance our requests about newborn screening with one from CDC that related to infectious disease screening in pregnant women that would have required checking of those results at, you know, labor and delivery in order to make sure or to minimize perinatal transfer. So they're trying to see if these two fit together or they're completely independent. But they are interested in pursuing a further discussion with the committee. And I'd be perfectly happy to

unload the discussion I've been having with them myself.

MALE SPEAKER: Is the recommendation, then, to have them as one of the stakeholders we're identifying as part of this debate?

DR. WATSON: I think they have a place in implementation to make sure certain things are happening and can certainly tighten a couple of loops, independent of just hospital-based activities or nursery-based activities.

DR. BOTKIN: Good. Well, we're going to have some general discussion now.

Tracy, I'm going to turn to you in a second.

And one of the questions I wanted to expose for the group, perhaps just to think about for a minute, are there other stakeholders that we've not identified as part of this process this morning that we need to be talking to in this larger debate. So I want to welcome any particular input about that question here in a minute.

Tracy?

DR. TROTTER: Yes, thank you. As a practicing pediatrician, I wanted to echo and clarify what a number of my colleagues said this morning, because there's two basic issues here that I think we're grappling with in terms of the focus of this last hour and-a-half. And Tim and Fred very nicely clarified the clinical practice issues. Both Chris and

Alan give away their backgrounds that they're really pediatricians underneath whatever other roles they have because they talk like pediatricians.

DR. FLEISCHMAN: We take that as a compliment.

(Laughter.)

DR. TROTTER: That's a compliment. Yeah, that is the kindest thing I could possibly say.

(Laughter.)

DR. TROTTER: So the idea of mandate versus clinical guidelines is absolutely critical for us to get our hand around. A well-appearing newborn with a time-critical issue that has a serious, life-threatening illness is a public health problem, I believe, and is a mandated issue, I believe. And maybe we won't all agree with that. But if you look at clinical guidelines, we talked about 50 percent. I think 50 percent's a generous thought, that we're all doing 50 percent of everything we're supposed to do.

Clinical guidelines are purposely written for wiggle room. I've written a few myself. And the opening paragraph is always, within the clinical context of making a decision with this individual patient, which is important for us many times. But I'd like to pose this question o the committee, to the room, everybody's here. Which of the current

recommended uniform panel tests would the average primary care clinician choose to screen for if it was an ala carte basis? All right? Every day, what would you? What would you pick off?

Would you say, well, I think I'm going to do this one because it's really rare, or there's no family history? Or are we going to get to go down that whole road? It's just that's why we do it that way. So we have to decide, does this raise itself to that level, whatever it is, whether it's -- I'm sort of thinking about heart disease. But there's going to be a lot of other things that come up.

Does it raise it to that level? If it's clinical guideline level, that's different. There is often no structured -- or if it's even structured, a poorly-organized, sort of, non-comprehensive follow-up, often by third-party payers or by the state or by somebody who may give me a point or not point on some profile I have. But that doesn't protect the patient, who I decide, you know, I don't think you need to have that today. But they actually did need to have that today. And there's no organized way to measure my success on a 100 percent basis all the time and increase the quality. That's number one.

Number two is I think Fred makes a very good point, that we have to be thinking outside the box a little bit about some of these recommendations we're coming up with regarding implementation.

Newborn screening traditionally is one of the, literally, most successful public health programs ever as a blood-based newborn dried blood spot-based testing. And we all know it. We're all comfortable with it, et cetera, et cetera.

And now as we go to some other thing that doesn't fit that, maybe it doesn't fit that. So maybe there is another public health entity, there's another way for this to be taken as opposed to trying to be shoved into a slot that's already up to its ears in very major problems keeping the rest of their programs going. That's not to say that it isn't a public health program. I'm not saying that. I'm saying it maybe is not a public health laboratory program in the way that we thought about it.

DR. BOTKIN: Thank you.

We'll stick with the committee here for just a minute and then, Ann, turn to you in a second.

Carol?

DR. GREENE: I'd like to maybe build slightly. Completely agree with what Tracy just said -- and build slightly and ask the question. I think people are making a very clear rationale for a mandate. And I think a lot of the discussion is, does it have to be a core newborn treating condition. Do we have to redefine newborn screening? Or can this committee -- and this committee is not restricted to newborn screening. If

we agree that the evidence is that this should be mandated, by what mechanism?

And if Mike hadn't brought it up, I was going to raise JCHAO, because they have fairly -- you know, if JCHAO says, thou shalt and you get down-checked, and you don't get your nursery recertified if you don't do it. There are other mechanisms that I think could be explored. So my question is, do we have to redefine newborn screening? Or can we recognize that this committee doesn't have to be restricted to newborn screening and say that it's clear this should be done and explore how?

DR. BOTKIN: Alan?

DR. FLEISCHMAN: Jeff, I just wanted to address the question of other stakeholders. I think the nursing community ought to be a stakeholder at the table. The Association of Women's Health, Obstetric and Neonatal Nurses in general represents that community that's in the nursery and at the bedside. And I think because this is such a test, that their wisdom and their support in terms of quality and implementation would be helpful.

DR. BOTKIN: Okay. Excellent.

Sharon?

DR. TERRY: Along the lines of other stakeholders, too, I would say the public. And I know that there are many publics. And so,

probably some combination of parents as well as advocates or a balance. I think some of the perspectives we just heard are very real. And it is a matter of policy we have to, as a nation, continually figure out how do we advance health, given restricted budgets and the kinds of decisions we have to make around economics.

DR. BOTKIN: Good. And we may want to be getting back to what's more guidance about that recommendation.

Chris?

DR. KUS: Yeah, I'd like to revisit that idea, redefining newborn screening. It was my impression -- I think the chair also recognized it, when we were talking about this, we included newborn hearing screenings. So, in my mind, it's already redefined. It's just that we haven't marketed that definition.

DR. BOTKIN: Don?

DR. BAILEY: This may be obvious to everyone already. But it seems to me that we're -- and I really appreciate the complexity of the discussion and the implementation issues, especially. It seems like we've been talking about anticipating new conditions that might be screened in the hospital as opposed to in the lab. And just want to make the comment that we ought to be anticipating in the future and asking the question about, well, what if the old conditions could be screened that way.

What if new technology came along, the PKU, for example, was best screened at a point of care in the hospital? I do think that day is probably coming eventually. And so, as we get to the process of evaluating these new conditions, we might want to keep that in mind and say, okay, well, what if this was PKU we were talking about. What would it mean? Or, and how would we would we be changing what we're doing?

DR. HOWELL: I think everybody's aware of the fact that there are current research programs looking at hospital-based screening for the things that we currently screen for in the lab.

DR. BOTKIN: Ann?

MS. COMEAU: Thank you. Is this on? Thank you. As a public health newborn screener, I wanted to offer a bit of a different perspective than what Fred offered, though I do respect all the budgetary comments that were made. And I wanted to draw upon what both Alan and Jane brought forward, which is you have to stick to the principles of newborn screening. And all of this discussion assumes that you have a valid laboratory test. And, to my knowledge, there is no other laboratory testing or population-based laboratory testing that undergoes such stringent adherence to quality assurance guidelines as newborn screening.

Certainly, in our newborn screening laboratory, if we have

multiple machines running, then we have to be able to correlate the reports from the multiple machines that are running in order to validate that the reports we're sending out on the kids are similar.

Lead-based screening -- you get a little bit more variety there. Much of the lead-based screening comes to a central laboratory. You might have some hospitals doing lead-based screening. But I don't think there's much -- I don't know enough to know how much correlation there is between the results that come out of lead-based screening.

If you're talking about point of care screening and you have hundreds of hospitals running hundreds of different instruments with a variety of different levels of people who are trained. One needs a very stringent quality assurance to prove the validity of the testing that's going forward if you're going to mandate this. And you already have a laboratory system in place that understands what you need for population-based screening. And I just wanted to offer that I think that the newborn screening programs do have a potential role, accepting the fact that different states might want to implement it in different ways. Thank you.

DR. BOTKIN: Thank you. We're officially out of time, so I want to look to Dr. Howell to see whether we should wrap up now or whether we have a little bit more time.

DR. HOWELL: You should wrap up briskly.

DR. BOTKIN: Briskly?

DR. HOWELL: Briskly.

DR. BOTKIN: That's the word of the day.

(Laughter.)

DR. BOTKIN: Yeah, I just want to make a comment in response to Ann. I agree with a lot of what she says. But remember how we got to newborn screening in the first place with PKU? Because all these different hospitals were doing different tests, and labs were doing different tests, and the results were a disaster? So the laboratories don't have control over that. And just by saying, okay, we're the ones to do it because we have experience in quality control does not make you the performer of the test. Thank you.

MS. COMEAU: We do collect a lot of data. I mean, you and I -- and we collect a lot of follow-up data. And we report it back as a quality control measure to people who are performing these things.

DR. HOWELL: Jeff, I think that Michele would like to make a comment.

DR. BOTKIN: Oh, please.

DR. HOWELL: Michele?

DR. LLOYD-PURYEAR: The few types of recommendations -- I'm just looking at the legislation -- that the committee can make -- and

this may be -- we should think about it, that -- or the committee should think about it. And this may address the newborn screening laboratories' concerns versus a newborn screening program's concerns, and maybe some of the health care professionals.

And I'm not a lawyer, so you guys know that. Anyway, one is --

DR. HOWELL: But she always carry the law that we work under with her.

DR. LLOYD-PURYEAR: One is making recommendations for the recommended uniform screening panel. And those are specific to newborn screening, or to newborns. And I don't know if it's just newborns, but it does say newborns in the legislation in the same place that it talks about the recommended uniform screening panel. Elsewhere, the legislative charge, besides just the name of the committee, is broader, where it either includes newborns and children or heritable disorders in general.

But under six, where it says, "provide such recommendations, advice or information as may be necessary to enhance, expand, improve the ability of the Secretary to reduce the mortality or morbidity from heritable disorders, which may include recommendations, advice or information dealing with," and one part is

conditions not included in the recommended uniform screening panel that are treatable with FDA-approved products or other safe and effective treatments as determined by scientific evidence and peer review.

And then, so that may apply or allow two sets, at least with these two things, this two sets of language for two different kinds of recommendation. One, that the committee recommends to the recommended uniform screening panel, which has immediate implications for the Affordable Care Act, in some states, more than Indiana, I think. And then other recommendations that you may want a newborn to be screened for, but you are not necessarily including in the recommended uniform screening panel. Again, I'm not a lawyer. So --

DR. HOWELL: We would not want to get into the position that we have an important condition like the congenital heart disease that escaped the ability to be covered under the insurance. We certainly would not want that to happen.

FEMALE SPEAKER: That's what I want. I wanted clarity on that last point, which is is the ACA, relative to HRSA, is just for the uniform panel. Is that correct? Does it actually say that in the ACA? I don't have it in front of me.

FEMALE SPEAKER: It's in the regulations that were written by the department that include Bright Futures. And Rod read them, and

we can read them again. Bright Futures --

FEMALE SPEAKER: They're in the briefing books.

FEMALE SPEAKER: Yeah. So do you want to --

MALE SPEAKER: It includes the core conditions.

FEMALE SPEAKER: It includes the core conditions. The recommended uniform screening panel recommended by this committee.

DR. HOWELL: And we would not want to exclude some of the important conditions from that recommendation. That would be my position.

We need to --

DR. BOTKIN: Yes. So I want to thank all of the speakers who have been willing to provide some insights for us today. I think we've made some excellent progress. And it's pretty clear we've got a lot of work to do at a pretty high altitude level, as Nancy suggested, with respect to the role of the committee itself with respect to a variety of different screening modalities and how best to collaborate with a variety of other stakeholders out there that are interested in the health and welfare of children.

So if folks have additional comments that they haven't had an opportunity to offer us today, please send those to Coleen or myself, and we'll incorporate those in the ongoing discussions at our

subcommittee level. And we'll have more discussion from the subcommittee during the course of this meeting and bring back to the main committee some recommendations about how to further pursue this complicated set of issues. Thank you.

DR. HOWELL: I would like to have -- thank you very much, Jeff and Coleen. That was an excellent discussion.

The committee has historically looked at newborn screening much broader than just a dried blood spot. Obviously, we have had hearing recommended for a long time. And I think that we should continue along that line. That's a personal opinion. And I think we'll see even more conditions. So I think it's important to figure out how these might fit into the program and so forth.

We are at the point of public comment. And I don't want to reduce that more than we already have done here. But we have, under the public comment area, on your thumb drive, you have two letters that are considered for public comment. One, the first of which, is from Assemblyman Jason O'Donnell of the New Jersey State Assembly. Assemblyman O'Donnell introduced legislation requiring that pulse oximetry be performed on every newborn in New Jersey who is at least 24 hours of age. That bill has passed unanimously in both the General Assembly and the State Senate and is now awaiting the governor's

signature.

Assemblyman O'Donnell's letter, obviously, supports newborn screening for severe congenital heart disease. And it also provides an account of his family's personal experience of having an infant diagnosed with congenital heart disease.

The second letter that's in your folder is from Dr. Emil Kakkas of the Kakkas Every Life Foundation. And Emil's comments are related to the session that we will have this afternoon on FDA policies and procedures relevant to individuals with rare, heritable disorders. He proposes in his letter revision to FDA policy that addresses emergency drug shortages that sometime occur in children being treated with rare diseases. And obviously, we will have an outstanding discussion by our colleagues from the FDA after lunch.

Our first presenter who signed up to give public comment is Jill Levy-Fisch from the Save the Babies Through Screening Foundation.

Jill, would you please come to the microphone and make your comments?

MS. LEVY-FISCH: Thank you for the opportunity to speak today. I'm going to be presenting to the Educational Subcommittee, but I wanted to make everybody aware. Save Babies Through Screening Foundation is in the final production phase of producing an educational

newborn screening video. We're about a week away now from hitting the focus groups before we have our final narration done.

We've shot film throughout the United States. Our footage is of diverse families and medical professionals. We have footage of screening lab facilities, multiple family interviews and innovative heel sticks currently being piloted in certain hospitals. Our medical professionals provide clear explanations on film of genetic disorders and the screening process.

They also cover vital information regarding positive screens and follow-up visits. In the video, families who have had their lives changed by newborn screening are interviewed and share their stories. In contrast, interviews with the parents of families whose child's hereditary disorder was not detected will also be shown. Engaging video of healthy children whose lives were saved by newborn screening will effectively illustrate the importance of the program.

We've worked with several experts who have been interviewed, such as Michele Hall, who is the newborn screening coordinator at UMC, Nevada, Brad Thorell, Dr. Melissa Wasserstein from Mount Sinai. We also are producing a segment for the Newborn Channel, which they're going to start airing in June, which is the four-minute piece. It's an educational film that'll be shown in 3,000 hospitals throughout the

United States. The channel will also be promoting their video -- the video in their journal and on their Web site. And this includes the distribution of our brochure to 6,000 health care facilities and health care providers in the country.

A longer version will be available for viewing on our Web site and on the Newborn Channel Web site. The video will also be posted to YouTube and available for linking by interested parties. DVDs are going to be distributed without charge to birthing classes, prenatal health care providers, nursing associations, et cetera.

We've worked with a wonderful, creative team. Our writer/producer is a multiple Emmy award winning news and feature journalist, a member of both the Writers and the Directors Guild and has produced and written for every major network. Our editor is a feature film editor who is currently working on the upcoming Disney feature, "The Odd Life of Timothy Green," with Jennifer Garner.

Our creative director is a design professional with experience in awards in multiple mediums. We've had wonderful advisers to the film, who I do have to thank: Dr. Maria Escolar, Michele Hall, Dr. Harry Hannen, Dr. Rod Howell, Brad Thorell, Kate Bockley, Melissa Wasserstein. And I just want to thank everybody for the time. And I'm hoping everybody will learn more about the video. We're going to be

showing it in two weeks at the IMAX summit and putting it out to the focus groups at that time. Thank you.

DR. HOWELL: And, Jill, I gather these videos will be available on your Web site?

MS. LEVY-FISCH: They'll be on the Web site. And some of the states have already expressed interest in linking to them. And the Newborn Channel is helping us promote everything, both the brochure and the video. And I forgot to add that everything will be done in English and Spanish.

DR. HOWELL: Thank you very much. And Annamarie Saarinen was to comment also. And she had some information that she would like to share with the group today.

MS. SAARINEN: That sounds great, Jill. I look forward to seeing that. Unfortunately, my comments grew a little bit because you put the comments after the presentation on broader point of care screening. But that was an excellent presentation.

I'll just start with, of course, the reason, primary reason I come to all of these meetings. And that is, of course, because of my daughter's diagnosis with critical congenital heart disease. And so, I've been following, of course, raptly everything that's been happening and was fortunately, like Chairman Hall, able to get the note with 90 minutes

advance notice on that phone call and able to listen in.

So I will tell you that a 90-day delay was a bummer, 1,000 babies that are going to die from critical congenital heart disease in 90 days. But in terms of federal process, I realize that's close to the speed of sound. And I think as best that this interagency coordinating committee can leverage the work that's been done by this fine group and the implementation work group, they will be in an excellent position to help address some of the Secretary's additional concerns.

I hope they can do more than just read through packets on paper and that there's some sort of face-to-face integration and involvement and picking up the phone and whatever needs to happen. Because without that level of collaboration, I can't imagine in 90 days them having an action plan that's going to really measurably move the ball forward.

With that said, this is all process. And the announcement from the Secretary probably could have been worse than what it was. So we'll take it for what it is, and I'm an optimist.

Dr. Vockley's comments on official response to the process -- one potential official response is Congress. And something I decided to embark on along with the Newborn Coalition and folks, parents and families of -- was to reach out to key members of Congress, the Senate

Health Committee in particular, Congressional Quality Care Coalition, the entire House Ways and Means Committee, so folks that actually have retained dialogue with the Secretary and weigh in really commending her historical support of all things related to newborn and child health and encouraging her to look closely at this important recommendation and that we're all eager to go ahead and implement and make sure children are not diagnosed late with heart disease, to the degree that we can, with pulse oximetry screening.

So a legislative update, quickly -- I just want to share this with you because what was said earlier about the guidance that comes out of this committee impacting what happens in the state level is absolutely true. A year-and-a-half ago, or more than that now, when, sort of, was considering this process and started going to the State Department of Health and then came to this committee, it seemed absolutely like the proper path and the right way to pursue things to adopt something as a standard of care, but also something that has received this level of guidance. And the states have been, sort of, waiting, knowing this is in process. But they're kind of not waiting any more.

So, as Dr. Hall said, the state of New Jersey passed legislation that sailed through the House and the Senate unanimously. I just got a note this morning that they have every expectation that the

governor will be signing that within days, perhaps even today. So Maryland, along with -- which also has passed their Houses, has been waiting for this guidance. They have questions and calls that have happened over the last two weeks, so that they can move forward and change their language slightly, actually, based on the sort of interim recommendation from the Secretary.

And four additional states have legislation that's been introduced, is making its way through committee. Four is already on the floor. And I think there's nine additional states with pilots or programs that are operating right now, either that have influenced the standard of care or are in pilot phase. There's about four or five additional states right now that have draft language. It just hasn't been introduced in committee yet.

So, I guess, my point is the world is moving forward with this with or without this committee and with or without the Secretary. I think the world is a better place with this recommendation because what's happening now, very quickly, in New Jersey is they've got 90 days to start implementing. The providers do. And without the sort of systematic guidance and, in theory, a toolkit that may come out of the work that's been done here, they're going to have to, kind of, reinvent the wheel a little bit to address the concerns there.

And that's not even really getting into the reporting and

follow-up issues that you've raised, Fred. So I think, you know, the closer we can get to a modeled solution, the easier it will be for hospitals and newborn nurseries to implement this in the most effective and efficient way. I think that's all I've got.

Oh, one more thing. Just this week in Minnesota, two patients this week in Minnesota, 15-month-old misdiagnosis, large VSD. They fortunately caught it in time now to do a surgery. But multiple, multiple trips to the hospital and to pediatricians, upper respiratory problems, every diagnosis other than congenital heart disease. So this happens. It gets missed by caregivers routinely. A five-week-old lived with half a heart, HLHS, five weeks old. They finally diagnosed it. She passed after two immediate surgeries as soon as they caught it. This is the kind of thing that pulse oximetry is really going to help with. It's not going to solve all the problems. It's really going to help.

So I commend you all for your work again. Really, I am just inspired by how far this has come. I hope this last little hurdle is just a hurdle we can push it over.

DR. HOWELL: Thank you very much. I think that the work that this committee has been done is certainly public. And we hope that the folks that are implementing it will take advantage of what has been learned and the -- as I pointed out, the workshop on implementation is

currently in press as a policy of the American Academy of Pediatrics. And hopefully, this will be helpful to the states as we move along to a final recommendation, hopefully, from the Secretary soon and so forth.

I think that that was the only folks that I had signed up on our list and so forth. And so, I think that that means we will leave a little bit early for lunch. Let me comment one thing, is that at our meetings here in the past, the restaurant upstairs has been overwhelmed with people. And so, the folks that are eating did not get back until two o'clock or so. And so, at the registration desk, they've got a list of other places in the immediate vicinity that might be able to serve you a bit more promptly. Because I do want you all back at one o'clock because we have a really terrific presentation from the FDA. Thank you very much.

(Whereupon, the meeting was recessed for lunch at 11:55 a.m.)