DACHDNC Webinar Conference
Day One
Transcript

Most participants are in listen-only mode except for those individuals who will be speaking. Conference leader, you may begin.

Joseph Bocchini:
Thank you. Good morning, everyone. I want to welcome you to the fourth meeting of the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children. I want to thank you all for participating in this newest iteration of our meeting. Hopefully, we have solved some of the problems of getting more people in the same room so that we could have further and better discussions about issues that are being vetted.

As you know, this is still a webinar format, so there are a number of people who are on the phone. We welcome you as well, even though you are not here.

This meeting has a full agenda. Our focus this morning will be on the public health impact analysis and how we can enhance it in order to inform the committee's decision-making process. Later today, we will hear from the CDC regarding the impact of electronic health record implementation on early hearing detection and intervention programs.

Tomorrow we will be discussing pilot studies, new CPT codes established for molecular diagnostics and their impact on genetic testing laboratories and patient access, as well as provide an update on the timeliness of the newborn screening project.

Before we begin, I want to announce a change in leadership of two of our subcommittees. The follow-up and treatment subcommittee, Carol Greene has done an exemplary job of leading this committee. Carol, I want to thank you for the work that you have done as chair of this subcommittee. As of September of this year, Charlie Homer, committee member will be taking over as chair of that subcommittee.

For the education and training subcommittee, Don Bailey has also done an excellent job as chair. As of September he will step down from that position and Cathy Wicklund has agreed to take over as chair. I want to again thank the current chairs for all the work that they've done and the products from those subcommittees, and I want to wish good luck to the new chairs as they begin their terms at running these committees.

Speaking of subcommittees, there are two reports there in the briefing book that need committee feedback. So please review these and make sure we are prepared to do that tomorrow. For follow-up and treatment, there is the framework for assessing outcomes for newborn screening on the road to measuring as promised, and from the laboratory standards and procedures committee, the succinylacetone as primary marker to detect tyrosemia type 1 in newborns and its measurement by newborn screening programs. So we'd like to have feedback from the committee to finalize those two products. And we'll do that tomorrow.

Next I want to give you an update on the reauthorization of the Newborn Screenings Saves Lives Act. The House Energy and Commerce Committee passed the Newborn Screenings Saves Lives Reauthorization Act of 2014, with only modest changes to the version the Health Subcommittee and Energy and Commerce agreed to back in February. The bill proposes to amend the Public Health Service Act to extend and improve programs at the Department of Health and Human Services related to newborn screening and it reauthorizes the advisory committee. So that is the status at the present time.

I would now like to turn this over to Debi Sarkar who will give us some reminders and housekeeping items. Debi.

Debi Sarkar:
Hello, everyone. Good morning. Just a couple of reminders and housekeeping items. You have heard these reminders before. So for the committee members, please remember that special government employees are prohibited from lobbying. So we cannot lobby as individuals or as committee, but you can in your professional capacity and as a private citizen. Also, interaction with the media or in the public, interaction regarding questions about the committee or if you're asked for an interview, please let me know beforehand.

For the people on the webinar, members of the public, you have two options in listening to the committee proceeding. You can either just listen via the Adobe connect. The sound will come through your speakers, or you can call in. There is a number on the left side of the screen. You can call in and hear it through your phone.

I've been asked by the USP building management to tell everyone that they are having other meetings in the building and those meetings will have food and drinks provided. Unfortunately, we are not able to have those food and drinks. But we do have a map of the closest restaurants and cafés where you can get drinks, coffee, and food. Also, there is water and water coolers.

Lastly, please have your badge with you at all times. It will help you get in and out of the meeting. Thank you very much. I'm really glad that we were all able to come together this time. It's been a long time since we've had everyone around the table. I thank you all for coming. I will turn it over back to Dr. Bocchini.

Joseph Bocchini:
All right. Thank you, Debi. I want to thank Debi and HRSA for their work to bring us together in this place. I think it's going to create an opportunity for a much better meeting.

Now I need to take roll call for this session. We do have three members of the committee who are unable to attend this meeting. One is Don Bailey, the second is

Charlie Homer, and then Alexis Thompson. Let's take a roll call of the rest of the committee. We will go around. Jeff Botkin.

Jeff Botkin: Here

Joseph Bocchini: Coleen Boyle

Coleen Boyle: Here

Joseph Bocchini: And AHRQ doesn't have a representative here at the moment. The FDA, Kellie Kelm.

Kellie Kelm: Here.

Joseph Bocchini: Fred Lorey?

Fred Lorey: Here

Joseph Bocchini: Michael Lu?

Michael Lu: Here.

Joseph Bocchini: Steve McDonough?

Steve McDonough: Here.

Joseph Bocchini: Dieter Matern?

Dieter Matern: Here.

Joseph Bocchini: Melissa Parisi?

Melissa Parisi: Here.

Joseph Bocchini: Cathy Wicklund?

Cathy Wicklund: Here.

Joseph Bocchini: Andrea Williams?

Andrea Williams: Here.

Joseph Bocchini: And our DFO Debi Sarkar.

Debi Sarkar: Here.

Joseph Bocchini
And I am here as well.

Organizational representatives. We'll take attendance of them as well. Representing American Academy of Family Physicians, Freddie Chen.

Freddie Chen: Here.

Joseph Bocchini: Representing the American Academy of Pediatrics, Beth Tarini.

Beth Tarini: Here.

Joseph Bocchini: American College of Medical Genetics, Michael Watson.

[Inaudible response]

Joseph Bocchini: American College of Obstetricians and Gynecologists, Nancy Rose.

Nancy Rose: Here.

Joseph Bocchini: Association of Maternal and Child Health Programs, Debbie Badawi.

Debbie Badawi: Here.

Joseph Bocchini: Association of Public Health Laboratories, Susan Tanksley. You should be on the phone.

Susan Tanksley: I'm on the phone. Can you hear me, Dr. Bocchini?

Joseph Bocchini: I can, Susan, thank you. Association of State and Territorial Health Officials, Chris Kus.

Operator: If you are on the phone, please signal me by pressing star zero.

Joseph Bocchini: Department of Defense, Adam Kanis. He should be on the phone as well.

Adam Kanis: Here via phone.

Joseph Bocchini: Thank you.

Genetic Alliance, Natasha Bonhomme?

Natasha Bonhomme: Here.

Joseph Bocchini: March of Dimes, Siobhan Dolan.

Siobhan Dolan: Here.

Joseph Bocchini: National Society of Genetic Counselors, Cate Walsh Vockley.

Cate Walsh Vockley: Here.

Joseph Bocchini: Society of Inherited Metabolic Disorders, Carol Greene.

Carol Greene: Here.

Joseph Bocchini
All right. Thank you all for your participation. Next we have on the agenda approval of the January 2014 meeting Minutes. These Minutes were included in your briefing document. Are there any additions or corrections to be made to the Minutes of the January 2014 meeting?

[Inaudible]

Joseph Bocchini
Great. We will accept those. Additional -- hearing none, we need to vote to accept the Minutes with the minor changes as indicated by Dr. Matern. So for the approval of January 2014 Minutes, Jeff Botkin, yes or no?

Jeff Botkin: Yes.

Joseph Bocchini: Coleen Boyle?

[Inaudible]

Joseph Bocchini: AHRQ not here. FDA, Kellie Kelm?

Kellie Kelm: Yes.

Joseph Bocchini: Fred Lorey?

Fred Lorey: Yes.

Joseph Bocchini: Michael Lu?

Michael Lu: Yes.

Joseph Bocchini: Steve McDonough?

Steve McDonough: Yes.

Joseph Bocchini: Dieter?

Dieter Matern: Yes.

Joseph Bocchini: Melissa Parisi?

Melissa Parisi: Yes.

Joseph Bocchini: Cathy Wicklund?

Cathy Wicklund: Yes.

Joseph Bocchini: Andrea Williams?

Andrea Williams: Yes.

Joseph Bocchini

And I vote yes.

All right. Thank you. This next discussion is an important one. This is public health impact. Can we put my slides up first?

All right. I want to give you some background information of how we've come to this point in the assessment of the public health impact. Just as a quick review. Some of the key responsibilities of the Discretionary Advisory Committee include the three that are on this slide. One is to make systematic evidence-based peer-reviewed recommendations, the most appropriate application of universal newborn screening tests. This includes technologies, policies, guidelines, and standards. The second was to develop a model decision matrix for newborn screening expansion, and that includes an evaluation of the public health impacts of that expansion. Then the third is to consider ways to ensure that all states are able to attain the capacity for screening short-term and long-term follow-up.

To go back to 2010 and 2011, when the committee made the decision to recommend to the Secretary routine screening in addition to the RUSP for critical congenital heart disease, the Secretary did accept that recommendation, but she reminded the committee about its requirement to determine public health impact with the following statement, that the recommendation was to collaborate with HRSA to complete a thorough evaluation of potential public health impact of universal screening for CCHD as required by our authorizing statute of the Public Health Service Act. Based on that,

the committee made some changes in its matrix for evaluation of the condition that comes before the committee for consideration for the RUSP. I think all of you are very familiar with this matrix. We added feasibility and readiness to the matrix and as we discussed this made the decision that public health impact could influence the outcome of the evaluation of net benefit but also and more importantly, could influence the feasibility and readiness of evaluation and that would then have an impact on the decision that was made by the committee.

Going forward, when we reviewed the nomination for Pompe, we did expand the public health impact assessment and we did so by including an assessment of the population level benefits using decision analysis, and this was done by the condition workgroup. And then we did include with the efforts of APHL a survey of the newborn screening program directors, followed by interviews with representatives of newborn screening programs to assess feasibility and readiness. As you know, the committee voted to include Pompe on the RUSP. That recommendation went to the Secretary. The Secretary has referred this to the interagency coordinating committee with the expected response at the end of July this year.

Following the Pompe decision, we had feedback from within the committee and from some stakeholders that the public health impact analysis needed to be strengthened. So that based on that, we had a number of discussions with state public health people, other organizations' representatives, HRSA. And we decided that we needed to focus on strengthening our public health impact evaluation. And we needed to do that before we made further decisions about the next two nominated conditions that we have accepted for conditions for the workgroup review. But we made the decision that we would move forward with understanding the public health impact analysis by identifying those elements that were necessary, and we delayed the deliberations on MPS 1 and ALD, and I made the committee aware of my decision to do that through an e-mail back in March.

With the help of a lot of people and including a lot of work from HRSA and most importantly I want to recognize APHL and Jelili Ojodu for the effort to put together an expert advisory panel meeting. They did so in a very short period of time. And that I think has been helpful to us to identify what we need to strengthen our public health impact assessment. We had this expert advisory panel meeting in April. It was held at APHL headquarters. The purpose of the meeting was to strengthen the public health impact assessment by developing a systematic approach for evaluation of all necessary information. I have listed here the members of the steering committee. We brought back Ned Calonge who was a member of this committee and has been very involved with a number of organizations on evidence review, and certainly a recognized leader in this area, to help us in developing this advisory panel meeting. Participants included committee members, stakeholders, and other experts. And this is a list of the participants. We did our best to try and bring in people who represented all of the stakeholders involved in newborn screening, ethicist, in addition, health economist, brought in the NCC and regional collaboratives, consumers, federal agencies were

represented. And then we also brought in evidence review experts to help us in making the decisions that we need to going forward.

So the overall results of that meeting was that it was felt our current matrix does not need refinement. The committee agrees, everybody would be comfortable with that. The key elements of public health impact I believe have been identified. And you did receive a summary of that meeting in your package briefing document. Today we will have a presentation to the committee for discussion and input into that document and the next steps will be sending the updated draft to the committee to the participants of the meeting, the regional collaboratives, for further input and feedback so we can refine it in such a way that we can then go forward and utilize it for the MPS and ADL reviews.

So with that, I would like to then turn this over to Alex Kemper and Jelili Ojodu, who will present a summary of the meeting and then we will open this up for discussion from the committee and then the rest of the group.

To introduce Dr. Kemper, Dr. Kemper is a general pediatrician, Director of the Program on Health Services Research at Duke University. His research focuses on the implementation and evaluation of screening programs for children including newborn screening, screening for visual impairment, and screening for lead poisoning. Dr. Kemper is also associate editor for pediatrics, the Official Journal of the American Academy of Pediatrics. He now leads the condition review workgroup.

I will also introduce Jelili Ojodu. Jelili is Director of the Newborn Screening and Genetics Program at the Association of Public Health Laboratories. He is also the Project Director for the Newborn Screening Technical Assistance and Evaluation Programs, NewSTEPs Program. Mr. Ojodu is responsible for providing guidance and direction for the Newborn Screening and Genetics in Public Health Program. He received his Master's in Public Health from George Washington University and a Bachelor of Science degree in biologic sciences from University of Maryland, College Park. So first, Dr. Kemper.

Alex Kemper:
Before I get going, I want to [inaudible - no audio]

Hello?

It's amazing. I've already learned today that an on switch makes it go on.

I do want to especially thank Jelili Ojodu for the work that APHL did in pulling this meeting together. And certainly K.K. Lam who works with me at Duke on this. So if you can go ahead and pull my slides up.

Fantastic. We have two hours on the schedule. What I would like to do is summarize what went on during the meeting. As I present these slides, if you have any clarifying questions, please go ahead and interrupt me. There are a lot of questions that we have

for the members of the advisory committee, things that would really help us as we continue to flesh this out and finish it up.

Everyone has gotten a copy of a draft of the report. I would like to remind everyone that it is a little bit of an older draft just because we keep getting comments in and revising it, so it may be one or two iterations old. What our plan is, after the completion of this meeting, to go back and revise the documents again and send it out to everyone for their feedback and continue until we have something that everyone is happy with. I'm going to go ahead and summarize the meeting that we had back in April and sort of where our thoughts are about this public health system impact.

So the next two slides are just a list of all the members, participants in the meeting. Since we put together this slide, I have gotten a lot of feedback about how people want their institutional affiliation listed and that kind of thing. So that's not all reflected on these slides, but I want to let participants know that we have all of that updated in the manuscript that will be coming out.

So that you can see in this slide, we had members really representing the broad spectrum of individuals who could help us think through the public health system impact assessment, including those from the laboratory side, the regional collaboratives, ethics experts, we had experts who have done public health impact assessments in other domains, state public health departments, genetic counselors, condition-specific specialists, primary care providers, and patient and family advocates, and then we also had representatives from several federal agencies. I was really pleased at the input that we were able to get for this meeting. Again, I would like to thank the participants for spending their time, both at the meeting and afterwards, as we wrestled with the topics.

This describes the meeting approach. We had key stakeholder groups involved at the meeting and I described those folks a minute ago. We sent out a lot of preparatory materials so that participants could dive right in, including information about how the RUSP works, what has been going on in the world of newborn screening and descriptions of other models of review and decision-making both for genetic testing and population-based health interventions in general. There were a series of meeting facilitators. At the end of the day, the meeting focused on three things:

- What elements should be considered as part of the public health system impact assessment;
- Who should we elicit to get those elements; and
- How should the information be gathered within the time and resource limitations available to the condition review workgroup.

Remember, again, the goal is to be able to go from nomination to decision within a period of nine months or so.

Again, I want to highlight the decision matrix which Dr. Bocchini talked about a few minutes ago. There are the issues of the net benefit and certainty of net benefit, most of

which will come from the systematic evidence review and from the decision analytic modeling that is coordinated by Dr. Prosser and her colleagues at University of Michigan.

When we think about the public health system impact assessment, it focuses on the issues of feasibility and readiness, sort of the right-hand side of the matrix. One of the things that I want to talk to you all about today is how we can provide the information to you in a way that will allow you to make the determination of readiness and feasibility, and exactly how to really prepare that information for you in a way that is clear and understandable and that can allow you to make transparent recommendations to the Secretary about nominated conditions.

So again I discussed the meeting objectives which were looking at the what, the who, and the how, and to identify how things can be used in decision-making.

At the end of the meeting, we kind of grouped things into general key considerations for what the public health system impact should assess. Those include

- Newborn screening program organization.

I'm going to drill into these in subsequent slides.

- The ability to screen;
- Issues related to short-term follow-up;
- Issues related to long-term follow-up.

And then there were a series of cross-cutting considerations, which include those related to

- Data systems and information exchange;
- The direct costs of adding a condition to newborn screening;
- Opportunity costs; and then
- Issues related to leadership and motivation to adopt screening.

So I'm going to be talking about those key considerations that are in bold, but you will see that the ones below that are not bolded will appear in my discussion of the key considerations.

Let's take a step back and think about issues related to newborn screening program organization and how they might play into the public health system impact assessment. There are issues related to the authorization process for adding a new condition, and that needs to be understood before a condition can be added. There is the process for obtaining additional funds to expand newborn screening for the targeted condition. There's the role of public health in providing access, and within that I include coverage after a positive screen for diagnostic services or treatment services for the child or for

the family. And when I think about the family, I'm thinking about things like referral to genetic counseling.

And again, these issues are going to vary state-by-state, but I think it's important for the advisory committee to have a sense of this. Of course, the issues that I talked about in terms of the role of the state in managing positive newborn screens really expands to issues of the targeted condition, so if you identify the condition that you are looking for. But as this group well knows, there is also the issues of incidental findings or secondary conditions, carriers, genotypes of uncertain significance, and then late onset disease. So again I think it's important to understand what the role of public health is across all the different things that can result from newborn screening.

So within the newborn screening program, there's also contextual factors that I think the advisory committee would benefit from understanding, things like motivation for change in leadership and how things get done.

So let's think about the next broad category related to the ability to screen. When we think about the ability to screen, it really bifurcates into laboratory-run tests, when I'm talking about newborn screening program laboratory. For now that is Dried Blood Spots, but who knows what that's going to look like in the future, versus Point-of-Care testing. So throughout the meeting we really separately considered how these two types of testing might play out.

In terms of laboratory considerations

- Whether or not there's a validated screening method and a platform that can be easily adopted by newborn screening programs;
- There's the issue of timeliness, so there's the time to analyze the specimen and the time to report;
- Availability of quality control materials and standards;
- Whether or not we are talking about a new test versus an extension of a process that's already in place, like having -- adding something else to standard tandem aspect;
- What is needed in the way of equipment and supplies; and
- What is needed in regards to laboratory employees.

By that I mean not only just getting new FTEs, but whether or not there's new training that needs to occur. A lot of these laboratory considerations are the same regardless of whether or not we are talking about a state that runs all of their samples within the state versus states that partner and have a centralized regional laboratory. Of course, in that case some of the timeliness issues become more complicated.

In terms of the Point of Care considerations, a lot of it is the same in terms of having a validated screen, but also issues like the ability to incorporate screening into the patient flow, the need for training, issues related to equipment and supplies, and what is the

added burden in terms of the need for new employees for Point of Care tests that are added. Unless anybody has questions about that slide?

Yes, Dieter?

[Unintelligible question/comment]

Okay, so that's definitely something we will have to put in and that's a nuance that I'm going to have to ask you all to help me. I'm looking at Kellie as well because I know there are this issues of kits versus home group.

[Unintelligible] That's a state law issue, not a --

Alex Kemper: Not an FDA issue. But I guess what I'm wondering is whether or not we ought to better understand, too, what it takes for the FDA to approve these screen tests which is beyond my --

[Unintelligible question/comment]

All right. I can learn.

What did I want to say? I think the committee is important to make note of that, though, because maybe the committee should advise states on whether they really need to have in their laws or statutes stating that it must be an FDA approved kit.

Alex Kemper: That's helpful I think. Dr. Boyle, were you going to -

Coleen Boyle: Maybe on another point, point of care considerations, thinking about what is going on with CCHD, the validated screen is pretty complex because it's a validated screen algorithm, but I feel like that minimizes the issues here.

Alex Kemper: I agree with you. There are two components that we are talking about. One is the validated instrument. And this has certainly come up in the CCHD world in terms of what pulse oximeter can you reliably use, and I know there is a lot of discussion around that, mostly from the manufacturers actually. And then the second point is how do you use that in the algorithm to make a decision. For the purposes of this slide I sort of abbreviated all that together, but I think that is really, really critical because we cannot tell birthing centers to go out and screen if it's unclear how they go about doing it. Of course, one of the things that the advisory committee is going to have to grapple with is how much evidence about the test characteristics of the algorithm are sufficient to make a recommendation. Any other questions before I move on?

[Unintelligible question/comment] I would expect that quality control standards would be applicable to point of care as well, but it is not listed.

Alex Kemper: Yes, that is true. When you look at the point of care considerations, it really sort of draws down upon the other things, and we will have to make sure that is clear.

[Unidentified speaker]: Thank you. Just a quick note about central reporting on point of care results?

Alex Kemper: We're going to talk about this whole issue of data infrastructure because that's the secret sauce for a lot of this stuff, and it is what states are really required to do in terms of tracking as well. I'm sort of stealing my own thunder, but one of the big conversations at the meeting was really understanding what the role of the state public health departments in terms of providing care versus really just providing a way to track individuals. So this whole issue of having data systems and being able to use meaningful health information exchange is really, really critical.

[Unidentified speaker]: Before you get to the tracking reporting, needs the training and needs the new employees. Both at the level of the hospital where the point of care is happening and at the state health department because if the state health department becomes responsible for they quality, they then become responsible for teaching people how to do it right and monitoring it's done right. So it's training and employees not just at the hospital, but also at the state health department.

Alex Kemper: Why don't we go to the next slide, because I have -- so, let me before I get to that, because I have those things under short-term follow-up. There were so many things out there that I kind of boiled them down into key concepts just to kind of make it easier to follow. So things that we need to come up out of the public health system impact assessment would be a description of laboratory technology, evaluation of the resources that are needed to implement it, so things like employees' qualification and expertise, materials, the data system, the expected need for short-term follow-up. So this issue of all the categories we have are artificial and they bleed across, but at the point of laboratory testing, understanding what the anticipated need is in terms of the number of people who are going to need follow-up. The impact on the existing laboratory screening process, and then issues related to impacts on cost, including consideration of newborn screening fees, which, again, is going to be something that varies state-by-state. So there's a lot of stuff that we talked about falls under these major categories. I put these together as a way to try to help keep us, or at least me, focused.

I have a similar thing here for point of care testing, description of equipment needed and evaluation of the effort required to provide the screening and report the results and evaluation of the data system needs, descriptions of the roles of newborn screening programs, which is going to vary state-by-state, generally that is played out with the CCHD screening. The expected need for short-term follow-up and evaluation of the expected costs. Again, that summarizes a lot of the discussions that we've just had.

Let's talk about the short-term follow-up. In terms of follow-up, there needs to be a defined process or an algorithm and that is what I think Dr. Boyle and others were alluding to as well. And that is going to vary by whether or not we are talking about newborn screening laboratory tests versus point of care tests. And then the role of the public health personnel and, as Dr. Greene was talking about a second go, there's the issue of data infrastructure and the health information exchange that is needed to perform whatever the requirements are within the state. And then there is this issue of the availability and accessibility and cost of diagnostic testing or whatever the follow-up testing that is required after a positive screen, including the need for a specialist to be involved. We all know that there are some conditions where there is a positive newborn screen then there is a simple -- and I say simple, not meaning to discount how complex it is to do short-term follow-up, but it's based on a follow-up laboratory test. Then there are other conditions where in addition to further laboratory testing, individuals need to be followed by a specialist which may or may not be available within the state. So there's the issue of whether or not those kinds of services can be provided in the state or whether it needs to go out of state, whether or not it can build on existing collaborations or if new collaborations need to be developed, either for the testing or the specialist for the short-term follow-up.

Does that make sense?

It either makes perfect sense or makes no sense. That's one of the dangers when you don't get feedback but I'm going to assume it makes perfect sense.

And then for long-term follow-up, again, data systems issues Dr. Greene brought up a second ago is critically important. And the issue again is whether or not the role of the state is just monitoring what happens versus being involved in service delivery. But in any case data systems are needed to track all that stuff. There's issues of availability and accessibility of treatment, even if the state isn't primarily responsible for providing that. There's issues again for out-of-state services and then the need to follow-up those who had variance of unknown significance or who are carriers or are presymptomatic and who might have late onset disease. Depending upon the condition, there is a whole spectrum of individuals that might need follow-up and understanding what the implications are for the state public health systems, obviously important. And all the issues I talked about can have implications both for the newborn screening program itself, the public health system, more broadly as well as Title V programs. I'm sure there are other categories in here that could be affected by the long-term follow-up that I haven't mentioned. Any comments on what I just said?

Everyone's with me. Freddie?

Freddie Chen: Alex, you've touched several times on this dichotomy between the service delivery side and this sort of public health side, and I think it's working, but at least I want to hear some more about sort of your thoughts around that because the topic is public health system impact. It's not health system impact, nor is it public health

impact. It's public health system impact. And I think it's really a scope issue which is really a struggle for -

Alex Kemper: I'm so glad you brought that up. I would love to hear what people say. The feedback that we got at the meeting was that even if the state isn't primarily responsible for providing the treatment, oftentimes, the state is -- and again I'm just talking about the long-term follow-up. You can generalize this with the short-term follow-up. But even if the state isn't responsible for actually insuring that the care is provided, there are implications for the state in terms of cost and that kind of thing. And then the other issue, too, is the state public health department can't begin screening for a condition if there is no way to provide the follow-on services. So at the meeting we really kind of struggled with where that line is. I think the challenge for putting together the report for the advisory committee at the end of the day, too, is how can we sort this stuff out in a fairly constrained period of time. So I guess what I'd like to do is turn it back to the advisory committee and hear what you all have to say about that.

[Unidentified speaker]: Before we start the discussion. If you have a comment, if you could identify yourself every time for the transcripts and for the people on the webinar. Thank you.

Carol Greene: Dr. Carol Greene, not a member of the committee but a liaison. I was about to bring up exactly the same subject and an interest in the discussion and to contribute to the discussion would point out that this committee has in the past under the leadership of Colleen Driscoll, come up with a document that's behind long-term follow-up and with all the care that has been taken for those bullet points, I think one thing was lacking in those bullet points, without saying who is responsible is that we have got data systems to monitor versus service delivery, and we have availability and access to treatment, we kind of hint at it [unintelligible] but I think we just have to say you have to have treatment. That is part of the definition of long-term care, and that's just a complex discussion and not saying who is responsible, but as you pointed out, if there is no care there's no point in screening. And I believe that is not captured in those bullets.

Debbie Badawi: I'm Debbie Badawi. I'm also not a member of the committee but a liaison. I wanted to get back to the point of differentiating the health system from the public health system because I think everywhere from when we look at point of care screening to doing the screening to short-term follow-up to long-term follow-up, there is in my experience actually a larger burden on the public health system to do that follow-up and to assess readiness for screening, if it's point of care. For CCHD screening, it takes a lot of manpower to go hospital–by-hospital and try and do some assessment of what's available as opposed to if it is a laboratory-based test, asking your lab director what they can do. So point of care screening in terms of assessing and collecting information on both short-term and long-term follow-up is a little more staff-heavy than the laboratory testing. So I think that's something to keep in mind when we do the surveys for public health.

[Unidentified speaker]: That's a very good point.

Debi Sarkar: I'm sorry to interrupt. This is Debi. If you could all hold the microphone closer, people are having a hard time hearing. Thank you.

Alex Kemper: Chris, do you have a microphone? I can't tell. Oh you do? It looks like a Norelco shaver.

Chris Kus: Okay. I think this is a critical issue. One of the things is the idea of really talks about the integration of the public health system with the healthcare delivery system. And it gets complicated in the sense of when you say state responsibility, because there's a public health state responsibility to monitor this. We now have health insurance that includes public insurance and then states are responsible for health exchange, so I think we have got to play that out, but I would then talk about the integration of public health and healthcare delivery and then within that, how do you define those kind of shared responsibilities because they are shared responsibilities, too?

Ann Comeau: Ann Comeau from University of Massachusetts. I would like to harken back to some of our discussions in evidence review. And that no matter where we draw the line for where the responsibilities are, when it comes down to feasibility, so much of these discussions are going to be dependent on the case definition that the advisory committee hands down because if we don't have a clear case definition for what we are looking for, whether it be at the screening level or not, then that responsibility becomes spread. If we don't have a clear case definition of what we need for short-term follow-up, then the feasibility of whether or not someone has a diagnostic mode for the one-week-old or the three-week old, versus the six-month-old, that responsibility then becomes very large for the public health people who are following this. To my mind, so much of this depended from the very beginning on what is it that we are looking for, and then all of the other questions still fall out from that. But if we can't first address this case definition issue --

Alex Kemper: That brings up a good point that I would like to address. Which is how we envision this whole thing playing out. So I think what would have to happen is that the first round of evidence review would have to be completed along with the issues of case definition and those kinds of things. And then the work that Lisa Prosser does at the University of Michigan would have to follow on fairly quickly after that so that we can anticipate what would happen when you started doing population-based screening in terms of the number of cases that would be identified and that kind of thing. And then before Jelili and his colleagues at APHL begin gathering the data, we would have to have opportunities to educate respondents about what it is that they are answering. So that we need to be able to go to them and say, you know, this is what the proposed screening test is, and I would like to point out that Ann Comeau has done a great job along with Susan Tanksley of coming out with that algorithm for explaining that to

states. But this is what the screen test involves and this is what the expected outcome of it would be, these are the treatments that are available, these are the kinds of specialists that would need to be involved, this is what would be expected from generalists. Whatever the issues are, that way the respondents can understand what it is that they are answering about. Because I agree with you that we cannot solicit information from public health folks without – so the way we envision it, too, is that there would be a series of webinars and other documents and stuff like that so that people would really understand what it is that they are responding about, and what kind of cases there are and stuff like that. So I agree with the comments that both Chris and Carol made a bit ago about the need for follow-up care and how ideally, that would be integrated within the public health department.

The challenge is that not all states are so forward thinking. So we would need to find out from their perspective about how things would play out. Hopefully, over time, that those kinds of things would develop. But the work that Jelili is going to have to do through APHL is going to have to reflect what the current state is with the health departments and how they see that playing out. And it's just the reality that some states are going to do it in a more comprehensive manner and include more of the kind of care that we are talking about, and other states are just going to – and I say just, but I know that it's difficult, but serve as just connecting the pieces. Still the financial impact is going to be felt by states, regardless of how it plays out. I don't know if I explained that well. Did what I just say make sense? More or less. Okay. We will flesh it out more because I'm going to have some questions for you all in a minute. Does somebody else have a question?

Actually it's a nice buildup in terms of -- we discussed that there's this whole body of information that we want to get. And we want to be able to get it in a well circumscribed period of time. I talked before about how we could educate people, but these are the kinds of people that ideally we would be eliciting information from. So it includes newborn screening program directors, the folks in the lab, the State Department of Public Health Commissioners, who they themselves might not have the answers, but could at least direct things to whoever the key players are. There's laboratory and clinical specialists in whatever the particular condition it is. And then depending upon the condition, too, primary care providers might need to be sought out. Chris?

Chris Kus: When I'm looking at that, I think one of the things that I'm concerned about is that there are -- you would be looking at individual public health commissioners. What about there is an Association for State and Territorial Health Officials? And within that there are policy statements with regard to newborn screening, which should affect that same thing with primary care providers. There are the Academy of Pediatrics, Family Practice that put out statements about that, and Maternal and Child Health Programs, Association for Maternal and Child Health Programs. So I guess the question is, how do you – you've got bodies that supposedly create policies to direct what health departments are supposed to do. They vary -

Alex Kemper: Can I ask you about that? So that did come up in the conversation. And the debate that ensued around that is whether or not these bodies could speak on behalf of what actually happened in individual states. So I'm a proud member of the American Academy of Pediatrics, and God knows I've been involved with a lot of statements, but we know that just because there's a statement doesn't mean that it's actually how things play out. It would be a million times more efficient if we could go to those bodies and get their perspectives on things. But the debate around it was whether or not that actually reflected what happened with an individual state. What are your thoughts?

Chris Kus: A couple thoughts. The idea is that in some ways they should be encouraging states to do what the group of -- like as far as you talk about the Association of State and Territorial Health Officials, they are a group that is supposed to reflect the sense of that across the country. So in some sense, that's a good place -- one of the questions that would come up is it depends on who you go ask in the states and what their opinion is. How do you balance that? Well, like everything, it's probably more territorial -- [unintelligible - multiple speakers] to blended.

[Unidentified speaker]: That's a hugely important point, because in terms of efficiency, if we could reach out to ask related groups to identify respondents because we do want to -- one of the hidden subtexts in this conversation is we want to reach out to all states and not just a selected group. So this is something that should follow-up when I get to the more the how kind of thing. But if we all could partner with that so in terms of at least getting the message out and getting people to respond to things, I think that would be a huge benefit.

Jelili Ojodu: Jelili with APHL. [Indiscernible] actually brought that up and that is one of the things that we're going to look at in the future.

Fred Lorey: I realize this is a summary and I threw this comment out through e-mail, but I think it's really important that we specify when we say clinical specialists in the condition that includes clinical specialists involved in the newborn screening systems because they're the ones that feel they've been left out. They are the ones that are best able to assess the public health impact on their patients and their practice. So I think that needs to be in there somewhere. And I realize this is a summary, --

Alex Kemper: Right. So it's not for example just having metabolic and genetic specialists, but a metabolic and genetic specialist who is interested in providing follow-up care after a positive newborn screening for fill-in-the-blank.

Natasha Bonhomme: Natasha Bonhomme with Genetic Alliance. Similar to what Fred was saying, I understand that this is a summary and I understand that we are talking about the public health system, but at the end of the day, the systems are in place for the family and for the public. And we do need to figure out a way to get some type of feedback to see if it is working on that side and necessarily just from what a clinician is

thinking is going on with their patients, but from the patients themselves and from the families themselves. Again, I understand what you are talking about in terms of key stakeholders and things like that, but the system isn't just in place for public health's sake, it's for the families' sake as well. So it would be nice in terms of as we continue to discuss this to think about how do we get those families and those on the ground experiences into a catalog as well.

Alex Kemper: We talked about that. I sort of lumped a lot of that in terms of the contextual features in terms of motivation for doing things. I'm 100% with you there.

Fred Lorey: I totally agree with you 100%. I think my point was in the past we have had some of that, if you remember the vote on Pompe? The attack I got from a parent group. But what we have not had in the past is input from these metabolic specialists, for example, who are involved in newborn -- and they feel left out. They feel like they did not get to express their opinion. So 100% I agree with you, but we've had some of that and we probably need more but we have not had that other element. And they are angry. They are angry.

Cathy Wicklund: I agree with that. And I think one thing to think about is, for any group that may be feeling left out at different phases of the discussion, and I don't know if you're going to get to this towards the end, is kind of what is the messaging that is going to go out around this topic outside of just the people who are in the room there or and the people who are fortunate enough to be in the room here, but whether that other clinicians who are in the clinic right now and not listening to the webinar or other groups that are going to be in the future nominating conditions, just if you had any discussions about the actual messaging and the pared-down summary --

Alex Kemper: That's something that I will lead to the advisory committee. Our task in specific was to evaluate what would happen if the condition were added, what will be the impact on the public health system. So that sort of got beyond what we talked about at the meeting.

[Unidentified speaker]: Yes, I'm just going back a little bit because you asked us what we thought about the idea of the healthcare system versus the public health system. The fact that we are asking clinical specialists and pediatrics, that is the healthcare system, too, correct? I mean, the fact that we are going out after these people, which I think is important. I think it's hard to just separate those two, there is impact on the healthcare system that is important, clearly, that we need that input on. So I feel like just the slide in itself is telling us that we are going there to get that input.

Alex Kemper: That's strongly my bias, but if people think I'm wrong, I'm welcome to hear you. What a great question.

Thank you.

Here you come with another one.

Freddy Chen: AAFP. I really raised the question for your sanity because --

Alex Kemper: My therapist appreciates that.

Freddy Chen: Because it really is so broad. And for me this issue of public health system impacts was exemplified by sort of when we discuss a condition and we ask the question, well, can the public health labs actually do it or not, and how much more is it going to cost them and the state and can that actually happen? I felt like it was a separate discussion than is there a treatment for it or are there specialists in Denver that can handle this. I felt like to be able to differentiate the health system questions, some of which are long-term follow-up from the actual public health system, I personally am not sure that a family physician would be the person to say whether or not the Washington State Department of Health, what the impact on the public health labs in Washington State. So that's the differentiation I was trying to make. I think that, given this discussion, which I can only imagine is a small reflection of the discussion you had, it gets pretty far ranging.

Alex Kemper: It's kind of like if you have a string on a sweater and you start pulling it. I'm going to get to this eventually, but primary care providers for a lot of conditions for the public health system impact, there may be no role and no need to ask them. I just want to lay out all the potential issues. But for things like the CCHD screening where a lot of primary care providers are obviously the folk in newborn nurseries, then it becomes more important. So what we were hoping to do is lay out what all the issues are, but one of the challenges that I will pose to the advisory committee is when we go to do a particular topic we will need guidance on where your priorities lie because we cannot do everything. And the other thing is there needs to be a transparent method that the advisory committee uses to weight these various things in order to make a decision because at the end of the day, you are going to have to throw a dart and hit the matrix in the right spot and so we need to come up with the buzzword is multi-criteria in decision-making tool. And so I will get to that in a little bit. But you're absolutely right. Dr. Chen, as always.

Ned Calonge: Yes. I just wanted to know whether or not you can actually hear me or not so that's useful. The thing I would add though is that there needs to be -- the public health impact does involve primary care providers and the fact that a lot of these kids are their patients. So yes, they may end up in the realm of the clinical specialist, but they also might end up in the realm of, oh, they need follow up testing and I'm just telling you if it is a child that I'm taking care of, and things are happening to the infant, that are ran by the state health department and the Children's Hospital or some other clinical expertise, and I don't know anything about it, that is an area where I would like to -- that has impact on my practice, how I interact with my patients and so I would say, I think you need to be a little sensitive to those hand-off areas and even just letting folks know because one of the worst feelings in the world is to see a patient that something has

happened to and they are now in your office and you are clueless. So, Fred, I would add that issue about handoff and notification is something that you might want to think about, especially tests. I remember the original testing for cystic fibrosis had a lot of false positives and so we were actually seeing lots of children in our primary care clinics with positive tests that we would've -- we might've wanted to provide some feedback to the public health department about how to better do that.

Alex Kemper: That's a very good point. Cathy and then Nancy or Carol?

Cathy Wicklund: Is there a way to think about this in the sense that there's things that will be weighed more heavily in the criteria, but then also things that are important for us to identify that might be gaps that we identify or issues that can be addressed in other – maybe not - I don't know they can be addressed by this committee, but we are raising the visibility of maybe workforce issues or different things that other groups can utilize to advocate for improvement? I hate to stop at something and not be able to use the information that we have to promote a lot of the things that we see that need to be fixed.

Alex Kemper: It looks like I just paid Cathy Wicklund to say that. That was very well stated.

Carol Greene: I believe it will build on what Cathy Wicklund just said. Realizing there is the integration and public health system and the healthcare system and private -- not to forget that especially for the specialists, but I think also some aspects of primary care, it does circle back to the public health system in dollars in a real sense because especially when the volume is low, especially in states where if you are going to have availability of care for methylmalonic or propionic or cystic fibrosis, you have to have the right people and nutritionists don't support themselves -- I'm looking straight at Debbie Badawi and part of my funds for paying my people's salary come straight out of the Health Department, and that comes straight out of HRSA. And so it all circles back around and it's all connected. Even if you think it is the healthcare system, it is also who's paying for the child on state health insurance, but also who is paying for the salaries for the people that have to provide services that are necessary that do not get paid for fee-for-service.

Alex Kemper: That's a great point. Because I have so many specific questions, I'm going to ask for the group, I'm going to plow ahead through some of the slides because I really want to make sure we get some specific advice from the advisory committee and that I leave time for that.

Chris Kus gave us some good advice a few minutes ago about gathering the data. We have really been struggling with how to do this in a very time efficient manner. And if you think about it, things break down into this general newborn screening information so that there is some data that aren't specific to any particular condition and may be available already from existing work that is out there. Things like adding a process, the process for adding new conditions or individual state's obligations for short and long-

term follow-up and information around existing infrastructure and workflow. So hopefully, in the future and again some is dependent upon funding issues from HRSA, existing information that is out there in general so that it will not have to be gotten each time.

And then the second category is this condition-specific newborn screening, and some of the data can be obtained from published works that are out there. But, again, that is going to have to be used to inform the survey's work that we alluded to a minute ago.

What I tried to do is summarize the broad thinking that occurred in the meeting, but again, what I want the advisory committee to focus on is at the end of the day, the information that is provided for the public health system impact assessment has to guide the advisory committee recommendations. And this is really going to be specifically around issues of readiness and feasibility. So I'm putting the matrix up again just to make sure that everyone is thinking about it.

Again, members in this room already talked about how some of these public health system impact assessments, and I thank Cathy Wicklund for her comments as well. But in terms of an assessment of the feasibility and readiness, but beyond that, identification of where the gaps are and how things can, based on this report, be used to facilitate better screening processes. It can be used to help develop a roadmap for implementation for those conditions that are recommended to be added to the RUSP, and also specific recommendations around those gaps or conditions that, for whatever reason, lack feasibility or readiness. So I think that the report can really be used for these four things as well as I'm sure other things that individuals can think of.

So the challenge though is taking these reports and using it to make a decision about where in the matrix things end up. We were fortunate at the advisory committee to have someone who is -- she has a great French name that I will not slaughter in public but from EVIDEM, which is one of many processes out there. It's a multi-criteria, multi-perspective decision analysis. I just went ahead and just stole this slide from them. And the process they use in making recommendations is that, if you look where that orange circle is, after the data are synthesized, then different weights are applied to different aspects of the decision that needs to be made. And then with that weighting system, that can either be used directly to make a decision, or as it is more often used, to facilitate the conversation around whether or not something should be added.

So for example, for a particular newborn screening issue, issues of the specialty availability might weigh large in the minds of the advisory committee, or issues related to if it was some brand-new thing, some brand-new technology that would be added to newborn screening, the ability to bring that new technology and train people, but other things might be weighed less heavily.

Again, we talked about a million different aspects of the public health system impact. I think that the way I would imagine this kind of thing being used is when a condition is

nominated and we have gone through the process of evidence review, we would develop in partnership with the advisory committee a list of the public health system impact questions that would need to be answered falling into these different categories. Again, this would have to be tailored to the individual condition. Then once everyone is happy with how the questions are stated or the broad categories, I don't think you have to look at the particular questions, but at least the topics to be gathered, we can then go back to the advisory committee and help us think about what things are more important and what things are less important, what's really going to drive a decision and what's not going to drive a decision. That will allow us to do two things: one was when we go, and when I say we I'm only talking about Jelili and his colleagues at APHL that are really going to be tasked with something very difficult. When they go in this very limited time period to gather the data, they would know what to focus on. And then also when it comes back to present the report, those items could be highlighted. Now, it's not to say that the other issues will be discounted, but it just will allow focus and would allow, once the decision is made for people to understand how the decision got there.

Now, when we had the meeting back in April in addition to the person from EVIDEM we also had Cindy Hinton from CDC. So we talked about other ways that the CDC is now beginning to elicit preferences. And what I thought we could do to sort of test how this kind of waiting process would work, is that after this document is done and we've developed a way for it in terms of doing the public health system impact assessment that everyone is happy with, we can actually go back to the members that participated in the meeting and do kind of like a trial weighting of the different categories just to see if it would work in the real world. And then once that process is done, come back to the advisory committee and talk about how to firm up that process or come up with a Plan B. But one of the many things that keeps me up at night is we are going to have this complex report to present for the advisory committee, and the advisory committee needs to be able to be clear about how it is that they use that information to decide readiness and feasibility. And certainly there are a lot of different components that came into it. So I would like to solicit what the advisory committee thinks about that.

So this is a way I see things playing out. So the condition review workgroup identifies a list of potential questions, and these can be broken down into either general or condition-specific. And then the advisory committee makes recommendations about those questions tailored to each condition. Then the condition review workgroup develops a list of the questions and then through APHL, administers the survey. Then a report is developed outlining the final -- I'm sorry, I skipped ahead. Before the survey sent out, the condition review workgroup prepares a report of the final questions, and then relative weight, and then the questions are done and the report is generated. And then the advisory committee makes decisions regarding feasibility and readiness, guided upon the questions that the advisory committee has already approved, as well as the relative weighting.

And I mentioned before the timing so I don't feel like I need to go through it again, but there would need to be an initial evidence review presented to the advisory committee

before the advisory committee can make certain informed recommendations about the categories of questions to be asked.

Again, this process that I've outlined serves two purposes. One is it allows the public health system impact assessment to be streamlined so that it can be done to complete a vote in time. And the second is it allows for clear and transparent recommendations about readiness and feasibility based on the results of the report. Now, I have no doubt that there's lots of complications built in and this will not work out exactly as I anticipate, but I think in terms of looking forward at least for MPS1, I think that this process at least trialing it makes the most sense here.

Again, I already mentioned before, we need to finish the report that came out of the meeting and that is going to be informed by feedback from you all. And then we're going to pilot the development of public health system impact questions and weighting with the members, the participants in that meeting that we had before. And then we'll be able to finalize that report for the advisory committee, and then use the revised approach with MPS1. So that's how I see things playing out. I've gone through the slide so I can open it up again. Dr. Lorey?

Fred Lorey: I agree that it would slow down your process to have it within, but wasn't that one of the charges from the Secretary, that it had to be included in the evidence-based review and not -

Alex Kemper: So we have - the final vote will happen after all three components are done. So there is the systematic evidence review component, there is the modeling component that is done by the University of Michigan and then there's the public health system impact assessment. So there would be no voting or anything that would be done until all those components are done. But the public health system impact assessment can't be done until we have a way to inform people about the condition in general. So just again, the way I see the thing going is the condition would be assigned to the condition review workgroup, and then at the subsequent meeting, there would be a presentation of the interim condition or the interim systematic evidence review in the same way that we've done with opportunity for feedback from the advisory committee about what things to beef up and what things are less important. In preparation for that meeting, though, is when the public health system impact assessment questions would need to be developed. And then the idea would be then for all three components to be done at the subsequent meeting in time for the advisory committee to make a vote. But there needs to be at least some floor of systematic evidence review to inform everything downstream.

Fred Lorey: I understand. And that would be the answer to my next question which was that means there cannot be a vote at the end of evidence-based review?

Alex Kemper: Well, that would be up to the chair because that is outside of my domain. But it would be my hope that all the pieces would be in place.

Fred Lorey: I see that as Alex put it together that the initial evaluation that would then come back to the committee would enable the things that were talked about, case definition, what would be the impact in terms of false-positives or other findings for carriers and etc. And then that would help inform the survey that would go out to then give better information to the states so they can provide us with information then that would be more useful because it would contain all the elements in a good way.

Ned Calonge: I do want to point out that I understand there would be condition-specific PHSI questions and hopefully some general ones as well. My concern comes down to the weighting. I understand how that works and the dimensional decision-making. The issue is that somebody needs to be paying attention condition-by-condition to the variation in the weights of those different questions. Because I can see that variation leading to different decisions and you may want different weights for different conditions, but then it's not very consistent. There's an inconsistency if there's a large amount of variability of how you weight the answers to the different questions. And I just want to make sure you are aware of that and someone's going to be keeping track long-term.

Alex Kemper: I think you're exactly right. As the membership of the advisory committee changes too, if there is that kind of drift, it needs to be transparent about why that drift occurs. That's something – remember I said there were a lot of things that keep me up at night? That's one of them. I think there are methods developed and maybe Cindy can talk about this more or certainly we have learned them from evidence, at least understand where the weightings are coming from and trying to keep that drift from happening. The other thing is that I don't see this the advisory committee being so wedded to like, this has a weight of .3 and based on that, then the decision is going to X. I think that it is going to inform a conversation. I do not think it is going to be a simple mathematical exercise as it is with some of the other multi-criteria decision-making tools. So point well noted. I think Dr. Bocchini was first.

Joseph Bocchini: This was very thoughtful and very comprehensive. I guess I'm a little worried about the impact on the public health system by the impact assessment process. [Laughter]. Sort of two questions. One is, understanding that this will happen after some data is available on net benefit. Because you wouldn't want to go to the impact stage unless you have a fairly high degree of confidence that there's already been established net benefit. So you will have a certain model for how screening was done in certain context to develop the evidence about benefits. So will states be provided that model to say, to what extent can you do what Wisconsin did or what California did, in achieving a successful screening model? So that they'll have some reference rather than sort of a free-floating idea about if you were to do screening for this condition, how would you do it and do you have the resources? So that's sort of one question. And then states presumably could say, well, we can do it but we would have to change the model in a certain way.

Secondly, it seems to me we would be most interested in states that don't declare themselves ready. In other words, if a state says, yeah, we're ready, we can do that, do we need a whole lot more data? Do they need to prepare a 20-page report on this or are we going to take their word for their readiness? So it's sort of the locus of control filing about who determines whether a state is ready or not. And if a state says we're ready, are we going to second guess that, or do we accept that at face value? And again, perhaps the data is more helpful for those states that would say we're unprepared. And if you have 25 states that are unprepared, it would be very interesting to know in what ways are they unprepared? How can the system help support states to become prepared?

Alex Kemper: I think the question that you get to is the granularity of data as well. And it is not going to be just enough to say, oh, yeah, we are ready and this is feasible. I will say that from having talked to many state health departments, and I've said this on many occasions, they're kind of heroes and they like a big task. If you ask, can you do X? Most of them will say, oh, yeah, we can do X. So it is going to really be drilling down. So if somebody says they are ready, what does it really mean? And that's where the questions developed from this group are going to be really key. I think the bigger question that you have on the advisory committee, which is one that fortunately Jelili and I don't have to resolve, is the degree to which the role of this advisory committee is to push versus being pulled. So if people say there is clear evidence of benefit, but a lot of people are not ready, how much does the advisory committee push? Or do you wait until there's some certain percentage of people that are ready? We can provide you with some of the answers from the perspective from the states and all these other people, but we can't tell you how to weight that in terms of the final decision, which makes my job easier.

Coleen Boyle: All great discussion questions, and this is obviously a very complex issue. You know how found I am of the 3D matrix. Do we need to separate the net benefit part of it from the actual implementation aspect of it? And not have everything sort of meshed together in a matrix. For me, personally this is very complicated and this is an extremely complex issue. Both of them are not static issues. Obviously they'll both evolve. The evidence review will evolve. There will be more information added. There will be more clarity, some more [unintelligible] and the adaption or adoption of this will happen. I keep wrestling in my own mind, I know this was a topic of discussion in the April meeting, so I'm just wondering if you could address that a little, whoever was at the April meeting. The actual matrix, keeping that already blessed matrix as part of this decision process.

[Unidentified speaker]: I think overall as we discussed this back and forth a number of times as we developed this matrix. The feeling was that originally we would do it in two parts, that we would do the net benefit first and then the public health impact second. But I think after that discussion it became clear that they were so entwined that one might inform the other and therefore it was put into the single matrix. Although most of the evidence review would lead to the net benefit, there are certainly possibly things in

the public health impact that could change the overall net benefit outcome. So we wanted to make sure that they were being done in parallel. So that's I think based on understanding the elements better that were needed for public health impact didn't seem to change that. And that is why we sort of left it the way it was.

Anne Comeau: Back to your process for evaluating public health system impact, I have more of a comment than a question. I don't know if you wanted to put that back up. The process to some extent in light of evaluating consistencies and inconsistencies and recognizing that there is going to be different weighting, the process itself seems kind of in danger of becoming a circular argument of the same people talking to each other. So my comment is that so long as the advisory committee meetings can have a public process with easy routes for corrections about misinterpretations of data. Has this happened in the past? For easy input from the public, then I think this works. But if we have to go back to just telephone webinars or just the advisory committee talking to itself, then this process is pretty much the advisory committee talking to itself and its own committees. And if there is data that are accidentally misinterpreted, there is no easy way to say, hey, wait a minute, we need to think about this. I think this is a good process as long as there is an easy way to have public comment.

Nancy Green: Nancy Green from Columbia University. Can you put up the slide about the matrix? Because I think that this is really a marvelous step forward and I certainly appreciate the thoughtfulness and experience. But I guess I'm going to make an analogy and I don't mean to trivialize anything, so please don't interpret this, but I hear that you kind of have to have the Preakness as opposed to winning the Kentucky Derby. What I mean is, you need to sort check off acceptability and work all of the different levels of review and not just one area. I think that's kind of where this discussion is going. Or could there be a California Chrome where something is so compelling that you can kind of push away the relative weighting of the other aspects? That is sort of one side of the coin. And then the other is, given those considerations when we think about the level through the evidence review, that the committee wants to attribute and in particular those sort of more let's say milky areas where there's moderate certainty or modest benefit. I guess the question is -- I guess the committee might have to consider, is it worth it. So sort of both sides of that coin, and what – so what's this really detailed more complex assessment, weighted assessment? What the impact may be on where the committee even bifurcates as far as moving forward in the Preakness.

Coleen Boyle: I guess I would follow-up with what Nancy said, and to me it's a bit of a staged process because depending on where you fall you might take a little bit of a different strategy there. I guess that's how I am thinking about this.

[Unidentified speaker]: I think that's an important comment and I think that if in that initial screen the evidence is clear that there may not be net benefit, that may stop the process. But I think that the benefit of the initial screen coming back to the committee having an evaluation of that, I think the committee's response will help inform the next part of the process. So but I think that can happen and that would fit the matrix if the

committee decided that the data really showed low likelihood of benefit or a potential for harm, that could stop the process right there. So I think that can happen and it can happen in the way the matrix has been set up.

Marci Sontag: I'm Marci Sontag from NEWSteps and Colorado School of Public Health. I just want to build on this conversation about the matrix. The way I think it's going to be implemented, from what I understood from the April meeting, something that ended up in that A4 category, we said it's high significant net benefit. And yet let's imagine the test that was $10,000 to screen every baby. So that's not feasible. We're not going to be able to do that, so I would suspect the committee might say that doesn't work. We can't recommend this right now because it is not feasible.

But in the A1, 2, and 3, A1 is a test that maybe on tandem mass we can use the current methods already in place. We just need to read out a new analyte. We could implement that pretty quickly. Yes, let's move forward. We'll recommend to the Secretary. But A3, all the states are completely unprepared. They can't do it. And yet our experience with this, what happens is the committee will say yes, we think it should be recommended but let's say the states can have five to seven years, ten years. Whatever that time period is, we're going to give them more time to do it. And yet that same results will happen. It will end up on the RUSP. The Secretary will say, yes, I think this is important; let's add it to the RUSP, and then it's another things that's added to that list. Is there a way to consider that RUSP to say, states, this is newly added and it's giving them a window to implement? So worry about the effect the public then has. They look at the RUSP and say, why is the disease that my child has on the RUSP but my state doesn't screen for it? Even though we said, this is tough. It's going to take ten years to get everything in place to do it. So how do we temper that information that's on the RUSP so the public can understand a little bit more behind all of these important decisions? Because I think at the end of the day, once it has a significant net benefit, it's likely to get added to the RUSP. Not and then you all decide, oh, let's forward it to the Secretary but that's likely that the timing piece is also important. So it's understood beyond just this group of individuals.

[Unidentified speaker]: That is a good point. When we created those three areas, the A1 through A3, we did include a potential timeline, within which it would be likely that all states would be able to participate. I'm hoping, with a better informed decision related to the elements necessary to make that case, that we can do better in putting it in the right category or going down to an A4 because it's not feasible to do that. If the goal here is to try and get all the elements needed to do a better job [unintelligible] those categories. We did include for the A3 category that it might take five to seven years for all states to be able to do what is necessary to include this in their routine screening. I don't know whether that answers, but I understand what you're saying about the public pressure then to go forward as quickly as possible because it has been approved for the RUSP. I understand that it is going to be a dilemma, and I think it'll always be attention on those things that are more difficult to add in. If there is clear net benefit, this committee may end up pushing the envelope a bit, and so there's going to be attention that's going to

be created based on that. I think that's true. But the goal is to do our best to kind of work together so that's not a bad problem in most cases.

Michael Lu: This is Michael Lu with HRSA. To follow up on Marci's question, there was quite a bit of a discussion during the April meeting about in addition to the impact on both the clinical and public health system, quite a bit of discussion around cost benefit calculus at the population level. I was wondering if you could provide a summary of that and also where in the process would the cost benefit calculus be done and how that would help inform the matrix?

Alex Kemper: Thank you. You are exactly right that there was a lot of conversation around the cost. I will tell you I went into the meeting being a little suspicious about whether or not the cost could be obtained. One of the things that I learned from the laboratory folk that were there was in fact they could maybe not put a dollar amount on the cost to implement the new test but could certainly put what they would need in terms of new equipment and what they would need in terms of new employees and that kind of thing. We can make similar estimates for cost across the system. Areas where things get hard for us to put a value on within a short period of time is the opportunity cost within the state public health departments. For example if a recommendation is made to adopt to screening for a new condition that involves a large initial outlay to purchase new equipment and to train people and to expand the data system to be able to track individuals and stuff like that, that's going to have an impact on the state's ability to provide services for something else that might be of value. So understanding those trade-offs is going to be really difficult. I don't think we're going to be able to put a specific number value on that kind of trade-off, but what I hope that we can do within the questions that we develop is from the state health department leadership to be able to elicit some of that information at least in sort of a qualitative sort of way. But that's going to have to be something that we're going to pilot when MPS1 is done.

In terms of the cost benefit, there is also the other component that was done by Dr. Prosser and her colleagues at the University of Michigan, where we can take the existing information around what we know about the screening test as well as the condition to be able to at least predict the number of cases that could be identified and the number of positives overall, and depending upon the condition, what the outcome might be in terms of years added to life or whatever data are out there to be able to predict health changes. And again, what we've learned is that really varies by condition and condition.

So getting back to it, there is the potential cost effectiveness or cost utility or cost whatever metric is out there, data that can be gotten from the modeling. There's the data that we can elicit directly from state newborn screening programs about estimated costs for introducing laboratory tests. This is going to be a lot harder to do for point of care newborn screening. So when that comes down the pike, we'll just have to figure out how to address it. And then we'll be able to, I hope, get at least a qualitative sense of what this would mean in terms of the opportunity cost within states. So I left the

meeting being a lot more optimistic about being able to collect those data, but I think we'll have to learn after MPS1 to figure out the ability for us, within the very tight timeline to be able to estimate that with any degree of confidence. We'll figure it out, but that was a big discussion. It was funny, I had cost on many of my slides and I kept waiting for someone to kind of bite and say, is this the purview of the advisory committee to think about cost? I think it is. But I was surprised that there weren't more comments on it. I see Kathy Hassell was back there raising her hands, too. Before I hand it off [unintelligible] capture that. Okay.

Robert Ostrander: Robert Ostrander, Academy of Family Physicians and Follow Up and Treatment Subcommittee. I just wanted to maybe focus a little bit on the role of the primary care community as a stakeholder in this from a public health standpoint. I think if I put myself in the public health circle of the Venn diagram, I think that for the purposes of our statutory mandate, I think probably the best way to frame that is to look at the primary care situation and workforce as a resource, and then that it's part of the public health system, and try to focus the discussions with the primary care community as stakeholders in how these conditions affect and put demands on that resource and use it to its best advantage. This does not happen in a vacuum. There's a lot of stresses and strains on the primary care workforce now, and it's a huge public health issue in general with the Affordable Care Act, with all sorts of valuable and valueless mandates for use electronic healthcare records, medical home sorts of things, and whether or not a condition is going to be primarily under the care of the primary care doctor, my perception is that a whole lot of the work involved in coordinating that, producing the interactions with the insurance companies, being the depository of data and information, a lot of that work is going to fall to the primary care folks. As you look at the follow-up and treatment in the feasibility stuff, I think that's where your conversation with the primary care community should be, as a stakeholder you look forward to this, how is this going to play out with putting demands on the primary care workforce? What jobs are going to be the jobs of the state health department? What jobs are going to be the jobs of the primary care medical home? And how are they going to interact with each other? A lot of the perception of the primary care community is that every time one of these public health challenges comes up, whether it's with heritable diseases or otherwise, there is a lot of downstream dumping of the work involved, and it's after a while overwhelming, and there's just so much time, treasure, and talent to devote on. Thanks.

Alex Kemper: I think Dr. Hassell is first and Siobhan.

Kathy Hassell: It's perhaps a coincidence that the geographic western half of the U.S. is sitting in the corner over there representing public health. I'm not sure it's the western half of the world. [Laughter].

I would just relay to the committee as a participant in this process, thank you again for the opportunity, and admittedly as an adult specialist and someone very engaged in the public health system.

A small or perhaps major part of this was this is seen by public health as the opportunity to say no to a condition because despite compelling evidence or otherwise, it's not going to fly for whatever reason a state believes. Now it's their belief and they are looking for the voice and the opportunity to say that. And so I think, thinking practically, and human beings being who they are, if in A1 you are never going to say no, no matter what the state says. At least when you ask, put it in the right context. Well, we're going to say yes no matter what, but we want to hear what your problems are going to be when this comes to pass because if you keep asking a lot of questions that might be a burden, the state will perhaps step forward to answer it. Because they perceive the opportunity to say, please wait, or please put it in A3 and give us a timeline. But they may come to it with the thought that they can actually stop the process. If that is not true, the first or second or third time you don't in their minds respect what they have to say, they're not going to say anything anymore.

Alex Kemper: I appreciate that perspective, and I'll add a twist on just to see what you think. So the advisory committee in its great wisdom has separated the condition review from the process that leads to decision. So given that it's going to be the process that provides information to the advisory committee about making a decision, we're not going to be in the position of speaking on behalf of the advisory committee, but what we can do is, if we interview a bunch of states and they say, no, we can't do this, and the reason we can't do it is for A, B, and C, then that needs to be highlighted in the report. From that perspective, I've said this about evidence, and I shouldn't say it about this public health system impact, and I think Jelili will agree with me. Our job is to pick up the rock and let the worms crawl out. We need to make sure that the advisory committee understands both the benefits and harms, and all of the challenges. And so it'll be our job to make sure that that's how it comes to them. Dr. Dolan.

Siobhan Dolan: Thank you. We just wanted to bring up the issue of safety and quality which really weren't addressed specifically in the narrative on the public health impact report because, and I thought of it with Dr. Ostrander's comment, as this is just pushed into the hospital and people see it as additional work, what are the mechanisms and safeguards [unintelligible] for making sure that as in more hospital-based tests, that safety and quality improvement processes are integrated into the assessment and then the implementation evaluation.

Alex Kemper: And I would like to point out that the March of Dimes has really played an important role around quality and safety, and so those points are very pertinent. Melissa.

Melissa Parisi: I have a question about the process per se, and I hope this isn't putting the cart before the horse. But to those conditions that might fall in this A4 category where the timeframe after the entire evidence review and the public health system assessment has been completed, would there be an opportunity to readdress the public health impact for those conditions, assuming that the condition review workgroup that a lot of the other things were being positive in terms of readiness and significance with a

net benefit? I am just wondering if that would require a change in some of the procedures and the way this committee operates, and if someone has thought through that.

Joseph Bocchini: Yes. I think part of the responsibility of the committee I believe is that if we get to that level, that we make recommendations because this is - a condition has been nominated by a group. We've gone through evidence review, we're going to go back to that group with specific recommendations of what's needed to potentially move it from an A4 to an A1 or so on. So, yes, I think that that is part of the process.

Melissa Parisi: Without necessarily needing to start at square one again, is really what I'm -

Joseph Bocchini: Yes, that's very true, and I think we've done that before. We've gone back, even with the nomination and prioritization committee, saying you have all these elements, but you need this before we'll look at this again. We've chosen not to go forward and move with the condition evidence review, but we've asked for certain information for that to go forward. So I think this is possible for the same thing, so that they don't have to start from ground zero.

There is the idea that you are going to the health department. Who speaks for the health department when you're asking, can you do it? Because you talk about opportunity costs, and is that the health commissioner or what does that mean?

Chris Kus: I represent ASTHO so I would say that you have to have the leadership of the health department because they've got the whole perspective of it integrated with comments. That's what I would -

Alex Kemper: That's what we were thinking, and we will hopefully. Dieter.

Dieter Matern: Coming back to the cost issue and also the involvement of the primary care provider, I think you should not only look at the cost of implementing an assays, but you should also look at the cost of the follow-up, particularly the follow-up of positive results that turn out to be false positives. I think it is extremely important to pay attention to the performance of the SA. What is the false positive rate? What is the positive negative value? That is one of those things you would have a hard time coming by to get that information from the screening programs. We really have to push that, and at some point we have to define what is an acceptable false positive rate for an assay or for the condition that we're screening for. Because we're talking about this all the time and evidence review we talk about these issues but we never define what an acceptable false positive rate X really is. And to determine the cost, I appreciate that every hospital or health insurance pays differently, but at least you can go back to CMS and the median cost for whatever procedure and determine there for it. Also using the ACMG Act Sheets and algorithms where it determines which tests you should actually

award every specialist who'd be involved. You can figure out what the cost for every false positive or positive result are at least for the short term.

Ned Calonge: This is not a new question for the committee. I guess the only thing I would add, I agree with the issues about assessing false positives. The problem is having a uniform acceptable false positive rate because it will really depend on the condition and the ramifications of what happens and the costs of a false positive versus the cost, etc. of a false negative. So I think, keeping in mind that there will be condition-specific issues that the committee has to organize, I think looking at the ramifications of false positives is important. I am uncertain you can pick a uniform cutoff level.

Chris Kus: I would just follow up with the idea of making sure we are talking about the cost and what it takes for long-term follow-up and treatment with a system that hasn't supported that. It's adding more and more conditions and we don't do very well with long-term follow up and treatment with the conditions that we already are following. So that's a critical part. I think we just haven't had robust enough resources.

Carol Greene: I think related to what Chris just said, when figuring the cost, figure the cost also of the treatment. I remember being asked when we were doing tandem spec in Colorado what percentage of babies have Medicaid because we had to figure the cost of providing the care for the babies that would then be identified. So really thinking all the way down, the cost of the screen, the cost of the false positive, and also the cost of not doing the screening. Because one of the things that I've always found very, very interesting is you can show the cost savings of doing screening, spend a dollar, save ten. And the problems come back to what Chris has just pointed out is spend a dollar, save ten, but you might save ten in the schools. Or spend a dollar, save ten, and it's all form healthcare, and the answer is, that's great but I don't have a dollar to spend because it'll come from someplace else. So looking at the cost, once you start looking at the cost, once you start looking at the cost, I think you have to look at the cost all the way down and broadly, and think about the cost of not doing the screening as well.

Alex Kemper: We will certainly do our best. So what I want to caution everyone is remember we have a very, very tight timeline to get this stuff out. So we are going to need your help to figure out for each condition what are the things that are of highest priority.

Natasha Bonhomme: To go along with that, and again I don't know if this is a question for you or back to the committee. Have there been thoughts of looking at the nomination form to try to gather as much of this information ahead of time? I know that in terms of the public health piece and talking to [unintelligible] but in terms of hoping to pull this information together because like you said there is a lot and people are looking for a quick turnaround time on the whole nomination process.

Alex Kemper: I will tell you from the evidence review side, the nomination forms are pretty well filled out and they work well for launching us, but I don't know if there's any more consideration on this.

Joseph Bocchini: I will say it is under consideration. And because of the tight timeline and what we are adding to this, that is something that we really need to look at and we'll her a little bit more about that tomorrow. That was a very excellent point.

Alex Kemper: Any other questions?

Jeff Botkin: Just one thought and it has to do with the language here in our matrix, which really assumes under the A category that there is a fair degree of uniformity in the determinations being made across the state. So A3 says public health departments are unprepared for screening. Now obviously what's much more likely is that we will find 20 states are quite prepared, 15 states are development, and a number of states say there's no way we're going to be able to do this in the next five years or so. Do we need a little more discussion about what that means in terms of our ability to say, well, based on the majority of states, that's how we're characterizing the overall system here so we're moving forward? Or is there some minimum set of states who are unprepared that are simply going to have to live with a positive decision despite their expressed lack of preparedness?

Dieter Matern: I have the feeling that the states do not necessarily care so much what we say anyway, so maybe we should just provide – well, I mean, we know what New York is doing. We know that some states don't implement all the conditions necessarily or it takes a long term. But why don't we look for the states that do a great job in providing efficient and effective newborn screening? And provide the other states, make them aware of how they do it, how they have accomplished what they have done as an example? And then they can go back to their own system and see what they might change? Which is going to be probably impossible, but at least there is something that we can point out that there are states that are doing an excellent job and maybe that is an example that they should follow.

Alex Kemper: Sylvia, way out there in Hawaii.

Sylvia Au: States care very much what disorders get added to panel, what this advisory committee does because basically when things are on the RUSP, it becomes an unfunded federal mandate. We get sued just like you do. Ask Minnesota, ask Texas. I think one of the things from public health impact is I'm lucky I live in a state we're very progressive in our genetics newborn screening, but all these things that you're talking about have unequal impact on small states with less resources. So even if you have great states that can do everything, there's lot of other states that, even if they have the will and the power, they just don't have the population or the resources to do it.

Rani Singh: Rani Singh from the Southeast Region Collaborative. I want to echo what Dieter said. In our region we definitely found there was a lot of will, and they wanted timely information from other states who had accomplished to be able to share the information from them so they could implement. So just sharing the information in a timely effective manner I would echo that thought.

Beth Tarini: Beth Tarini, Liaison from the AAP. I also have the position as the chairwoman of the Quality Advisory and Assurance Committee for the State of Michigan, where that committee is responsible for making recommendation to the legislature on the disorders that will be added. So I can echo Sylvia's comment that we do take the RUSP decisions into serious consideration. And I also wanted to temper the use of the word who's doing a great job because a great job I would contend does not necessarily mean did you or did you not add the disorder at a given time? I would argue it's are you working diligently on the process? And the process as we all have heard everyone comment, requires a lot of resources and a lot of cost. And also as I've heard commented, each state has its own legislative and policy restrictions by which it can fund such endeavors. And not everyone can go to the trough for the newborn screening fee again and again. Some rely heavily also on general state funds and revenues which are, as everyone knows, not easy to come by. So I just want to temper the use of a qualitative value statement like that. The states can be working diligently and these are not easy resources to come by.

Fred Lorey: Actually I agree with everybody, Sylvia and Dieter and Beth. I think we are talking about two different uses of the word state, and not to speak for Dieter, but if you're looking at the context of legislative action from states, irrespective of input from the health departments but maybe from parents groups, support groups. It's true. They don't care what the committee says. We have seen it happen in state after state. They want to introduce something and they've got a group and they find an author, they do it. So two definitions for the word state. I think we're talking about, so everybody's right.

Joseph Bocchini: Jelili and Alex, thank you both very much for organizing this and getting us to where we are. I think our goal obviously is to develop a better understanding of the public health impact, and I think that we have come a long way with this with input from stakeholders to identify those elements and now we obviously have to work hard to try and make sure that we obtain them in an effective way and then utilize them as a committee. So what I'd like to do, at the end here it says ask the committee if there are additional things, if we're on the right track, if there are other things we need to be thinking about as we evolve this public health impact evaluation. Dieter.

Dieter Matern: One more question. In the document that was provided as well you indicated there were stakeholders from third party payers invited, three of those. One declined and two did not respond. Why not? Why don't they care?

Alex Kemper: There was a very short window from the time that the research became available to hold the meeting. The meeting was organized and they just physically could not come. But that does not mean they are not interested and it doesn't mean we're going to back and solicit advice from them. But that was the one group that was absent.

Joseph Bocchini: -- comments from the committee members? All right. Again, I want to thank everybody. This has been a very good discussion. I think we're moving in the right direction and I appreciate everybody's effort to bring us to where we are. So that will close this discussion. I think we will have the next iteration during the next draft soon and for everybody to look at and make comments on it.

The next item on the agenda is for public comments. I want to inform the individuals who have requested the opportunity to make public comments that we have time for each person to speak for three minutes. Operator, please stand by to open the lines.

Operator: Thank you. At this time, Joyce Wulf, if you are on the phone please signal me by pressing star zero.

-- [Silence]

We do have Dean Suhr available by phone.

Joseph Bocchini: Mr. Suhr, please go ahead.

Dean Suhr: Yes, good morning. Fascinating discussion on the public health session there. Greetings. Thank you, committee chair and committee members. This is a quick follow-up on a message that I've been conveying to you over the past few meetings with regards to convening a summit to discuss one of the committee's current criteria for approval of newborn screening which is the requirement for a viable therapy. As I've indicated in the past, we understand this is a complicated issue with a lot of different perspectives. And our goal is to reopen that discussion in a roundtable format, kind of a casual format to understand how those perspectives may have changed over time. And depending on the results of the roundtable, then provide feedback in to the committee. We understand that we do need structure as far as how things are approved for the RUSP, and we're not arbitrarily saying we'll take this away. We do believe there may be some changes in perspectives.

We have previously stated that we tried to get that roundtable scheduled in concert with your September meeting, but it looks like due to some scheduling and funding conflicts, that we're going to defer that until the January, 2015 meeting, and likely try and do that maybe the afternoon before for those members of the committee or the public that may want to sit in as well. So we'll keep you posted on that. Anybody that is interested, if they could please contact me. My email is *dean@mldfoundation.org*, and available on the web and so on.

Just a quick word to those advocates in the public that may be listening in, the Newborn Screening Saves Lives Act apparently was passed by the Senate back in late January but it remains stalled at the House's Energy and Commerce Health subcommittee. That bill number in the House is HR1281, and I encourage all of the advocacy groups to be aware of that and to let them know that the committee needs funding, that we need to continue on with the good work that all of you are doing. Thank you for your time and efforts.

Joseph Bocchini: Thank you for your comments, Mr. Suhr. We appreciate them. Next on our list is Joyce Wulf, parent advocate, Council for Bile Acid Deficiency Disease. Is Ms. Wulf on the phone?

Operator: This is the operator. Ms. Wulf has not signaled. Ms. Wulf, if you are on the phone, please signal me by pressing star zero so I can open up your line.

[Silence]

Joseph Bocchini: If she has not signaled, the next person on the list is Sandra LaPrad, Consumer Task Force Member, Baby's First Test/Genetic Alliance. She's actually here.

Sandra LaPrad: My name is Sandra LaPrad. I am a member of the 2014 Consumer Task Force for Newborn Screening. I want to say that my comments are coming from me, not necessarily the Genetic Alliance, although I do appreciate Natasha's comments about getting families involved because that's kind of my [unintelligible]. I'm the mom of two beautiful girls, age 12 and 13. My 13-year-old was diagnosed with PKU by newborn screening when she was eight weeks old. Having no knowledge of anyone diagnosed with a metabolic conditions, we researched the internet and found all kinds of misinformation about PKU including speech and gait problems, musty odor, mental delays and eczema. With our first clinic visit, we were reassured that these problems typically only occur with poorly or nontreated PKU. However we did not see another child with PKU or speak to another family with a PKU child for five long months. It was then and only then at our first PKU family get-together that we were reassured that our daughter could have a typical life despite her diet restrictions. We were able to see PKU kids and families interacting. Kids fighting with their siblings, running and jumping, eating low protein foods and taking formula. So connecting families together is crucial. Because PKU and the other diseases screened by newborn screening are rare, often this may be the only child in that county with a primary doctor who does not have any experience with the treatment of these disorders which is how metabolic and specialty centers come into play. The specialist will assume the care for the specific diseases and should be sharing notes with the primary doctor at the medical home. Parents should also be involved. They should be educated as to what some of the long-term challenges might be with a child with their specific disease. For example, a PKU child might have an executive function disorder or difficulty as this has fairly recently been discovered to

be a challenge even with the most compliant PKUer. Parents should be knowledgeable about how this would manifest itself in the child with that disorder and some treatment suggestions.

Parents can also help the medical professionals understand more about their child's condition by speaking with their local birthing hospitals, general prenatal educator, nurse midwife, nursing schools and med schools about newborn screening. Doing this type of presentation puts a personal touch to the professional. They can relate to the diseases by putting a face to it, making it more memorable. Informing groups about newborn screening should be the first step. What is it? Why is it important to have it done and done correctly? What are your state guidelines? How is it done? How are families notified? Questions are asked to the family during those discussions that may help the audience with possible future patients of their own. Topics such as daycare, how do you afford medical food and formula? How do you travel? All kinds of other day-to-day activities. How families manage when they have a non-typical child. For families wanting to help increase awareness, this type of interaction can be done by simply asking their specialist within their clinic or hospital to do this, or contacting a local university or community college. The possibilities are pretty much endless. Increasing awareness and knowledge of newborn screening is the key. Thank you.

Joseph Bocchini: Thank you very much. I echo the importance of family interaction, education and awareness. Next we have Sarah Wilkerson, parent advocate, Save Babies Through Screening Foundation. She's here.

Sarah Wilkerson: My name is Sarah Wilkerson. I'm here as a mother and as a member of the Board of the Save Babies Through Screening Foundation. My son Noah was featured in the *Milwaukee Journal Sentinel* series last fall for passing away a few days old from undiagnosed MCADD. He was born on the wrong day of the week in my home state of Colorado, since the lab was closed on the weekend, adding a few extra days to us learning of his illness. As you know, a lot has changed since the article series ran, and a number of positive changes are taking place. In terms of standardizing the policies and procedures with turning around newborn screening test results in a timely fashion. I'm closely following the Newborn Screening Saves Lives Reauthorization Act. I'm incredibly grateful for the added verbiage on this bill. I'm eager for it to pass and see the changes that will result. I also want to second the comments about giving support if you can. I'm also very interested and thankful of the work being done by Dr. McCabe and the March of Dimes with the consortium. I want to thank the organizations that are participating. I know many of them are represented here today on the committee. I also appreciate the work being done by that committee to refine best practices here as well. I want to thank everyone involved in researching the subject of timeliness. My only request is that in my own research after my son died, I found a number of best practice documents, many of which would address timeliness in some form or fashion. It seems that these documents tend to fall flat when it comes to enforcing them. They'll sit on a website after creation and maybe hit the right people in the labs and hospitals who may or may not choose to follow it, but I urge you not to stop at simply the completion of

such a document. Think carefully about how to ensure that this valuable resource gets into the right hands and stays front and center in their minds with the increasing demands of turnover, budget constraints, time constraints and so forth.

I'm also interested to hear updates on the committee's plan to approach the Joint Commission which would be a tremendous move forward if successful. Though that only impacts issues on the hospital side of things. In my own story, my son's death was caused mostly by the state lab since they were closed when we needed them the most. I would like to know what reciprocal steps could be made in cleaning up web policies and not just hospitals.

I want to leave you with a story of another family that I recently became connected with. Just like in my story, they lost their daughter Ellie at a few days old in June of 2013. What happened? She was born on a Friday in Massachusetts. In Massachusetts the state lab is closed on the weekends or may have limited functionality for a few hours on a Saturday if need be, but Ellie's test sample was delayed as a result and she died. Her illness was undiagnosed MCADD just like my son. These stories are continuing to happen across the country, and I implore you to keep pushing full steam ahead on this issue so that children like Ellie and my son Noah can live long and healthy lives that they're entitled to live. Thank you.

Joseph Bocchini: Thank you, Ms. Wilkerson. First of all, it's nice to meet you in person and have you here, so we appreciate you coming. And thank you for your efforts, and certainly it was your phone call to us that initiated our concerns about looking into this issue. You will hear tomorrow feedback about where we are in the evaluations. And thank you for coming.

Operator? Can we see if Joyce Wulf is now on the line?

Operator: Joyce Wulf is on the line. One moment please.

Joyce Wulf: Thank you all for what you are doing. I represent the parent advocacy group for the Council for Bile Acid Deficiency Disease or CBADD. I wish to make public comment urging the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children to directly establish screening for inborn errors of bile acid metabolism in newborns. There are nine often fatal congenital deficiency enzyme defects which involve bile acid synthesis affecting approximately 400 to 500 newborns a year. There is a subset of the approximately 4 million live births that numbers 20,000 newborns. It is this population that is often grouped together as having idiopathic cholestasis detectable by a simple common test, direct bilirubin, that is often not done. This group of 20,000 is that that should be screened for these often fatal inborn errors of bile acid metabolism. This efficiency screening would catch nearly all of the DNA mutations in bile acid deficiency disease, allowing life-saving treatment to begin immediately. The parent advocacy group of CBADD understands first-hand the dire consequences of late diagnosis of bile acid deficiency diseases. Undiagnosed and

untreated, our children have suffered from fat soluble vitamin deficiencies, failure to thrive, delayed growth, bouts of profuse perspiration and a sickening buildup of toxins in their little bodies. With treatment some of our children are living completely normal lives. Others have attained a far superior quality of life than living under the crippling effects of untreated bile acid deficiency and threat of early death. Where once our children's day began with jaundice, blindness, fragile bones, no appetite, danger of brain bleeds, crying, the kind of crying that nothing can comfort, now each day dawns there is laughter, learning, eating that doesn't sicken, running to play, and no more tears.

The United network for Organ Sharing lists 494 children between one and five awaiting liver transplants. Perhaps as high as 20% of these children would not need to be on the list if there was a newborn screening mandate for bile acid deficiency disorders. Bile acid deficiency disorders are treatable and the treatment is highly effective if begun right after birth. Seven to five years to wait from a parent's point of view is unacceptable when every day is critical. I encourage you as a committee to push the envelope. Thank you very much.

Joseph Bocchini: Thank you, Ms. Wulf, for bringing this to the attention of the committee. We are certainly happy to talk with you further about deficiency diseases and to hear additional information. So thank you for bringing this to our attention.

That will close our morning session. We now have an hour and 15 minutes for a lunch break. As Debi mentioned to you earlier, in your packet there is a list of local restaurants. So we will look forward to seeing you back here promptly so we can start at 1:15.

[Lunch break. The conference will resume at 1:15 Eastern time.]

Operator: If Debi Sarkar has called in, signal me by pressing the star and then zero. Thank you.

Good afternoon.

Joseph Bocchini: Let's go ahead and start with me, and then Coleen.

Coleen Boyle: I'm here.

Joseph Bocchini: And then representing AHRQ, we have Mia Desoto. Thank you. And then Kellie Kelm.

Kellie Kelm: Here.

Joseph Bocchini: Still waiting for Fred and Michael Lu, Steve McDonough. Joan Scott will be representing HRSA for Michael Lu.

Joan Scott: Here.

Joseph Bocchini: Dieter Matern.

Dieter Matern: Here.

Joseph Bocchini: We are waiting for Cathy Wicklund.

Cathy Wicklund: Here.

Joseph Bocchini: And Debi Sarkar.

Debi Sarkar: Here.

Joseph Bocchini: For the organizational representatives, Freddie Chen.

Freddie Chen: Here.

Joseph Bocchini: Beth Tarini.

Beth Tarini: Here.

Joseph Bocchini: Michael Watson.

Michael Watson: Here.

Joseph Bocchini: Nancy Rose isn't here yet. Debbie Badawi. Susan Tanksley by phone.

Susan Tanksley: I'm here.

Joseph Bocchini: Thank you, Susan. Chris Kus?

Chris Kus: Here.

Joseph Bocchini: Adam Kanis by phone.

Adam Kanis: Here.

Joseph Bocchini: Thank you. Natasha is not back yet. Siobhan Dolan.

Siobhan Dolan: Here.

Joseph Bocchini: Cate Walsh-Vockley.

Cate Walsh-Vockley: Here.

Joseph Bocchini: And Carol Greene.

Carol Greene: Here.

Joseph Bocchini: And Natasha --

Natasha Bonhomme: Here.

Joseph Bocchini: -- is here. And Andrea is now here. Okay. We'll see who comes in over the next few minutes. I just want to quickly inform attendees who drove here, if you were not aware of this, there is visitor parking available. [Break in tape] to our attendees, parking lot is located to the side of this building. The entrance to the visitor parking lot is located on Twinbrook Parkway. If you choose to use the visitor parking lot, please remember to sign in with your vehicle tag information at the registration desk located just outside the meeting room.

At the end of this presentation and discussion Debi will have some information about the subcommittee meetings and how we need to get organized for those.

So this afternoon we have a presentation on the impact of electronic health records implementation on the early hearing detection and intervention programs.

To present this information is John Eichwald, M.A. who is Branch Chief of the Child Development Disability Division of Human Development and Disability at the National Center or Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention. John Eichwald is a team lead for the early hearing detection and intervention program within the National Center of Birth Defects and Developmental Disabilities at the CDC in Atlanta. In this position he oversees various activities including collaboration with federal national and state agencies and organizations, and assisting the states and territories to develop standardized procedures for newborn hearing screening, tracking, surveillance, data collection and management. John has more than 30 years' experience in the field of pediatric audiology. He is published extensively and made numerous presentations primarily concerned with early identification of hearing loss in newborns. We are really happy to have you here today and looking forward to your presentation. Thank you.

Coleen Boyle: Can I - if I just a few minutes of introductions – thank you. When I mentioned to John, this was back last year probably about doing a presentation on our work on electronic health exchange of information, it was before I had the opportunity to actually attend a recent public health informatics conference held at CDC. At that conference there was actually a demonstration of exchange of health information across various systems. One of those demonstrations was around newborn screening, and

obviously because EHDI is out there in front, early hearing detection intervention, that was the demonstration.

It was actually quite eye-opening for me personally, because a lot of this is words that doesn't have a lot of meaning unless you actually see it in action. So it was taking information at the time of childbirth, the creation of electronic health record. That electronic health record information was transmitted to the state health department through an exchange, an electronic exchange of information. That information was then transmitted to the primary care provider, and then subsequently, and again it just depends on how rapid this whole process evolves in terms of people time. That information was then filtered on to the audiologists.

All of that happens in real time, and all that allows for the ability for us to track what is going on both from a clinical systems perspective, and from a public health system perspective. So a lot what we talked about earlier in terms of evaluation, metrics and understanding the timeliness of what is going on and being able to evaluate, I think what EHDI has done and hoping that the rest of newborn screening comes in behind that, it's just a great example. It still is very complex to get all of these systems to communicate well, but I think this is a great demonstration of that fact that it does work, and that this can be useful for us, because there is real-time data.

John Eichwald: Thank you Dr. Boyle. That's all we need to say. [Laughter]
Before I get into the presentation and particularly after some of the comments by parents today, I want to give you sort of a history of why I have a passion in the area. This was 40 years ago when I worked at Utah Department of Health and my job was as a graduate student was to call up parents who filled out a questionnaire in the hospital to find out if their child had a higher risk for hearing loss. My job was to call them up and say, hello, I'm John Eichwald. I'm calling from the health department. You had a questionnaire that you filled out in the hospital, and you should bring your child in for a hearing test.

And far too many times there would be a silence and Mom would tell me, I'm sorry; my baby died. Because of those kinds of things, I've had a personal mantra my entire professional career, and that is, we have to have the right information, at the right time, for the right person in the right format. And I keep following that, and I think electronic health records are one of those formats that we really ought to be keeping an eye on.

We have been doing this in collaboration at times with the advisory committee for about the last seven years. This is sort of a timeline of what's been going on in the organization that we've been working with. It starts back in the days of the American health informatics community, and then health information standards panel.

[Break in audio]

Thank you. And then we switched gears after the HITECH Act. HITECH is the Health Information Technology Economic and Clinical Health Act. That was part of the American Reinvestment Recovery Act, known as the stimulus package under the Obama Administration, and that put in nearly $20 billion into the systems for hospitals and providers to get incentives to use electronic health records and also establish the Office of the National Coordinator, or ONC.

There have been other organizations we've worked with, and I'm going to go into much greater detail with some of those. Continuing on with the history here. Back in 2008 with people here from the advisory committee, we developed a use case for newborn screening. This was a use case that followed both blood spots screening and newborn hearing screening. I know the details are too small for you to see here, but this talks about the whole system of following a child from screening, getting the child into diagnostics, and it was basically sort of a blueprint of what we want to do.

That was shortly thereafter followed by the interoperability specification, developed by the Health Information Technology Standards Panel, which basically took that use case, and said how can we start moving data around? This was just prior to some of the HITECH money getting into the system.

At about the same time again, we started working with Integrating the Healthcare Enterprise, that is IHE. IHE is a nonprofit organization which covers several domains including cardiology, ophthalmology, radiology. There is a committee within the IHE that's called quality research in public health. It was formed about this time to start looking at this kind of information. It's a group of people, vendors and organizations, quality measure people, joint commission on this, and a number of public health organizations, and CDC has a fairly good representation on this committee.

And so in 2010, we published one of the first profiles for newborn screening. This is a profile that was published. It did cover newborn screening that is both blood spots and hearing screening. That was followed later that year by a profile that we published specifically on newborn hearing screening. This is really sort of the time were EHDI sort of went off on its own and started pushing into this area.

I know we have a number of layers of expertise in the room and I'm going to be sort of switching from easy and also very technical. So after lunch I will bore you at least once.

I want to bring this down to a real simple idea here, and that's basic communication. We have information that we need to send and we have to have that information received. From a technology perspective, we talk about this as a content creator. There is three different interoperability definitions I'm going to talk about. This is semantic interoperability. And then as we have to be talking the same language, we have a number of standards that have been developed. We have ICD-9, CPTs. We've got LOINC codes which is logical observation indices names and codes. We've got SNOMED codes, that's a systemized nomenclature for medicine, clinical terminology.

We've got pharmacy codes. We've got radiology codes. We've got value sets and I'll talk a little bit more about value sets. For example how do we code race? There are several ways to code race and ethnicity. So this is for the first things.

How do we create that information? Hopefully this is recognizable to many of the people in the room. This came directly out of the committee. This is the newborn screening coding and terminology guide that was developed primarily by our colleagues at NIH. This was done several years ago, but you can tell that this is a more recent edition, because it also includes the coding for clinical congenital heart disease. This continues to be upgraded. Here you can see we've got a listing of those SNOMED codes, ICD codes, and you can link on any one of these and drill down to the coding systems and the semantics that we want to be transmitting.

All of this information is also contained by NIH in what is called the Value Set Authority Center or VSAC. If you look down here you can actually see that we can click down and get even deeper and deeper into some of these code sets, live birth, hearing screening codes. And then to make sure that everybody is on the same page, we also have another place where all this information is stored, and that is at PHIN VADS, and that's at CDC. And that's the Vocabulary Access and Distribution System. All of this information is the same information that's being provided to people in terms of standardization for newborn hearing screening.

We've got the standard that we know what we want to move. Then we talk to what is the kind of technical, how are we going to move information from one place to another? Primarily what we're looking at here, is working with HL7. This is an HL7 message. This is a version that's going to be considered one of the 2.5 – well, this has 2.5.1, but it's a version 2 HL7. I'm just going to talk sort of real quickly so you get an idea of what constitutes an HL7 message. This is just one small segment.

So that first paragraph basically is an order. You can see we've got these delimiters between them, these hats, or bars and these little carats are what are called hats. Those divide each one of these categories within that order. Once you get used to looking at these, I can tell that this is a newborn hearing screen panel that was ordered by Dr. Minnie Smiles. Then we go into the actual observations. So those next four is the information contained in an HL7 message about the screening.

If we look at the third paragraph there, you can see this is newborn hearing screening result for the right ear and it's pass. The next one is the newborn hearing screening for the left ear and it's a pass. The next observation at the bottom paragraph says that testing was done by auditory brainstem response. This is a standardized way that we can be moving information for this between systems.

I also want to talk about other work we're doing in HL7. This is the EHDI implementation guide. This is an implementation guide that has been published by HL7. It's for secure transmission from the screening equipment to an information systems so that we do not

have to have that data entered manually. That was published as a data standard for trial use, so it is basically out there now being tried within the systems.

And so this is the kind of information that would be contained within capturing data from a device, not only a device but also capturing from electronic health records so we've got screening results from device or the reason why the screening wasn't done, we can get some basic patient demographics. We can also capture risk indicators about late onset hearing loss if it exists within the system, how long the screening took, who the screener was, and some real details about the device itself.

And then when that message is sent, we can assure that it was sent without any errors and then we can get an acknowledgment the message was sent. This is done within an implementation guide.

This is just one table within the entire implementation guide. And this has sort of been fixed up a little bit because I did some cutting and pasting and I got rid of some tables or columns that didn't look, and change some rows around.

For example this is an implementation for an HL7 message for a patient identification. What's going to be down here in that bottom, so in that stream of data is going to include a patient identifier. And in the guidance say we can have up to four patient identifiers, and that would include medical record numbers, billing numbers, birth certificate identification or if a national ID existed.

Cardinality means we have to have one but we can have multiple. The optionality says that is a required field. This information has to be set. You cannot send that message without that identifier.

Patient name again required. Then the value set, patient name again, getting back to those value sets, those national coding systems we want to be using. The RE means it's required if it's available. If you have the data you must send it. The optionality here is you only send only one mother's maiden name, date of birth, only one date of birth. There is the actual format that's recommended. I think that is an HL7 format. Again, race, here is an HL7 value set for race and for ethnicity.

That was done for newborn hearing screening. That was the data standard for trial use for newborn hearing screening. At the same time, this was also published for critical congenital heart disease. The same standards or extremely similar standard has been developed for pulse oximetry to capture the information from the devices, that demographic information, and send it into, again, from a device in EHRs to public health.

We also worked with HL7 on a public health functional profile. This is guidance to electronic health record vendors, and that is what should be contained within an electronic health record for the purposes of public health. The first one was published in

2011 and we dealt with three systems. That was EHDI, Vital Records, and Cancer Surveillance.

What we looked at is what do we want to specifically have in electronic records for these public health programs? It consisted of a series of statements that an EHR shall, should or may have A, B, C, D, this is what we want in it.

Then in 2013 it was just republished and now we've added other public health domains including lab, health statistics, occupational disease, birth defects, DVT, PE and adverse events and hospital reporting. This document is essentially based around a spreadsheet. That spreadsheet now has 3,000 lines of information of shoulds, shalls, and mays. We have a column for each one of these domains. Just as an example one cell within one row within one of those, in the end column under EHDI, here is an example. That is here's the conformance criteria, that the system should provide the ability to capture and store newborn hearing screening from devices and transmit those results. That's again for publication.

Nothing we want to talk about in terms of HL7 is what's called clinical document architecture or CDA. This is for all practical purposes version 3 of HL7. There is some real nice things about this new way of messaging information. The HL7 message, if you remember, that was basically just a string of information, and it's passed through and it's captured and then it sort of disappears.

CDA has persistence; that is, we can continue to look at it. We also know where it was created. There's stewardship. We can authenticate who created this. There is full context clinically contained within it, in a very comprehensive way, it's for wholeness and a critical part in here, it's also sent a format that is human readable.

So this is just something that I wrote. It's based on XML, which is extensible markup language. So instead of that string of information, this is the coding that sits behind it. You will see on the left-hand column, this is the XML code, and that would contain all the information you're going to send according to the guidance you are provided. You as a receiver, can take that information and put it into what's called a style sheet, take whatever pieces of information you want to see, and put it out in a format that can be read by humans. That is the one thing that's really nice about CDA is it is both computer readable and human readable.

We have been testing this out. Last year we had two pilot projects. One we did in Oregon, one we've done in North Dakota. This basically shows when we did this in collaboration with the public health data standards consortium, we took information from the electronic health records, in this case it was Epic, and we sent that information over on what's called a continuity of care document which – we won't get into great detail here. The OZ system served as our health information exchange. That information, they took it, created this information and created a hearing screening document in the CDA format. Then that was consumed by the public health department in the State of

Oregon. They were able to capture this and put into their system which is based on FileMaker.

In North Dakota we did similar, but we sort of modified it a little bit. Here again, we were using Epic. In this case, information was sent as an ASCII file, again, sort of just a text file. It included capturing data from the device and some newborn demographics from the electronic record. That was sent to the public health program in North Dakota. And then what they did was they created a CDA document that was this early hearing care plan. And then that was electronically submitted to a pediatrician's office. And then we demonstrated that successfully in 2013.

I'm going to also touch just briefly on consolidated CDA. This is really sort of a standard that is being pushed forward, sort of that comprehensive record. This is that continuity of care of information that we want to send in electronic message. You can see here that there is information on the care plan that would've been sort of the care plan that we did in Oregon, but it also has lab results in here. It's got histories, procedures, and problems. So this is sort of the standard that will be used in the future. If you're going to submit an electronic record, move an electronic record, this will be the standard of the minimum kind of information that we want to send as part of a, for example as a discharge summary from a hospital.

So now we've talked about, now we're getting into the third part of interoperability or process interoperability. You have to consume be able to consume this message. Now we're moving into integrating the healthcare enterprise. Remember, there were a couple of early profiles we did. One for newborn screening and one for EHDI. We've developed a couple of other profiles. One of those is a quality measure for execution. This is a quality measure that I will talk about in a moment. Our early hearing care plan which is being revised into what's called a hearing plan of care, and then workflow document and I will touch briefly on these.

I need to back up because of that quality measure. In 2011 we worked with the National Quality Forum, and we had several quality measures that were endorsed by NQF. Specifically from the CDC we had hearing screening prior to discharge, audiological diagnosis before three months of age, and then intervention no later than six months of age, also the HRSA indirect measures, levels of proportion of infants covered by newborn blood spot screening, and then NCQA, newborn hearing screening. That one is a measure where we want to make sure the physician's electronic health record has hearing screening results. Those were all endorsed in 2011.

CMS picked up on this as part of the meaningful use in stage 2. They have now taken this one measure which is the NQF 1354, which is now CMS 31, hearing screening prior to hospital discharge. This is now one of 29 clinical quality measures that hospitals can use to get to qualify for the CMS incentive programs for meaningful use. So there are 29 out there and they have to choose 16. That does not mean that EHDI will be chosen, but there is three other newborn quality measures, so we are sort of hoping they going

to align. If they do one infant measure, they will do more. This actually went into effect October 1, 2013. Hospitals can now use this as part of their qualification to CMS for certified electronic health record.

We worked with Lantana Consulting, MITRE Corporation, and Oklahoma Foundation for Medical Quality to really refine this down to a measure that can be done strictly electronically. As I said CMS has picked up on it. To qualify the hospital only has to report one quarter's worth of data. I suspect everybody will wait for that last quarter to submit and so we are holding our breath to see what hospitals have picked up our measure and will be reporting this as part of their certification.

All of this is contained. AHRQ has actually taken all of these 29 clinical measures plus other objectives within CMS. If you really want to know the details about this measure, you can go out to USHIK, the US Health Information Knowledge Base. Again, you can drill down in here and look at the definitions for the numerator, the definitions for the denominator, what are those codes that we want to be using that a hospital can use? So this is the place I'd recommend if you really want to get more information specifically about our measure.

ONC, Office of National Coordinator, has published a number of health vendors right now that have been certified that can report clinical quality measures so you can see we're getting some pretty good uptake.

This is our latest profile that we are developing right now. This is our technical framework supplement. It's really a plan of care. It actually will be going out for public comment probably next week, no later than next Friday for 30 days. If you go to this website at the bottom you can actually get through there and read the profile. I think it is 140 pages long. Some of it is very technical, but some of it is very basic in terms of – that's the stuff I provide. It's not the geek stuff. I provide the basic information for hearing screening. We're really trying to do this on a jurisdictional basis so the jurisdiction can decide what goes into that care plan and provide that to physicians, provide this to audiologists, provide this to all the specialty services that we need.

This is a made-up screenshot from the hearing care plan. You can see once again what we're talking about is producing this at the CDA format and it is human readable. If you can look down here, we're capturing again race and ethnicity, patient identification information, information about the screening.

The actual plan of care, what happens depending on the results of the hearing screening results, what will be in that care plan? Does the child need follow-up? Does the child need to be seen by an audiologist? Do they need counseling on late onset hearing loss? This is basically within that we've got the risk indicators, procedures that were done, active problem list. And so this is what we're putting out right now for public comment.

I also want to talk about NANI. This is our Newborn Admission Notification Information System. We don't want to just take information and send it to one place. When data is created, you ought to be able to use it multiple times. We don't want to re-create the same information over and over again. So what has been developed within IHE is this profile called NANI. It basically automates that information, capturing the information from a birthing hospital. It basically comes out of the admission, discharge, and transfer information, but it sends a message to public health that a baby has been born. It also talks about – so this information will be sent to public health.

We are using it in places to create our denominator so we know every baby has been screened and we can use that for our quality measure. Now this specifically was being implemented for EHDI; however, it could be used for any of the public health programs, and that is anytime you want to know that a baby has been born and capture that information and send it. So we could be using this for blood spot. We could be using this for immunizations or anybody else who wants to know information about newborns.

The one thing that is really nice about NANI is it does not have to be installed in the hospital. The manager – so remember we've got that content creator consumer but somebody has to manage that. That can actually be installed either in the electronic health record itself, it can be installed in the Health Information Exchange, or it can actually be installed at public health, and it works in all three. Just so we know that when a baby is born, that public health is informed.

This is basically a snapshot of all of the things we have been doing. And demonstrating as Dr. Boyle talked about. We've got our NANI, we've got this messaging from devices, we're creating care plans and we're doing quality measurement all electronically.

We have been demonstrating this for several years. Primarily we have been demonstrating this at HIMSS showcases. We've also been demonstrating it at some public health conferences. The last one in the bottom right-hand corner is the one that Dr. Boyle was talking about where she saw it. It had been showing different profiles over these times, again showing that we can do this and we can work with multiple vendors. We can work with multiple health departments.

That is not the whole picture where we want to go, though. That is we don't want to just send information one way. We want to make sure that we're communicating or what's called bi-directional exchange of information.

We talked about an actor and a content creator and a content consumer. Well, that content consumer can also create its own information. Really what we are talking about is how can this information again be used multiple times in both directions? A system can consume information, it can take that information, re-create, add information to it, send it back to the original source. So we really want to have that right information at the right time and in a timely format. So that's really sort of the direction we are looking.

We have done some demonstration of this. This was the showcase we did in Orlando. They changed up the way we did demonstrations before this time. In this case what we did this year is actually took a patient and ran a patient through the system. And particularly the one that we were working on was patient number 2. This was a 35-week neonate, and so we showed how we could capture vital records information, create electronic birth certificates, discharge that child from hospital care to the pediatric unit, and capture hearing screening results.

These were the vendors. I just want to acknowledge their work that they did. You can see that we worked with a number of EHR vendors. We also were working with Health Information Exchange and in this case we worked with the Minnesota Department of Health to capture the birth certificate information.

This is the demonstration we did at the public informatics conference just this last April. And again what we're doing here, is again capturing that information. And again what we're doing here is we took information from electronic health records and created both electronic birth certificates and newborn screening information and sent to the public health.

These were the vendors that we used in Atlanta, and again I want to recognize them. In this case we are working with the Utah Department of Health, so they were creating their electronic birth certificates.

We're working with the Office of the National Coordinator on a number of initiatives, primarily – and it was actually yesterday at 4:00 o'clock was the final phone call for the public health reporting initiative. We are really trying to establish standards of information that we can capture in public health, structured data capture, what are those actual coding systems that we all want to agree upon that we can use that is common to all public health programs? We're also working with the clinical quality framework and that's quality measures that we can be capturing that way. All of these have been combined, the structured data capture, data access framework and healthy decisions which is clinical decision support, into a new initiative of the ONC office, Office of National Coordinator, called the public health Tiger team. We are now working on that. We're actually talking about doing a new demonstration as part of the Tiger team, where again, we're going to be showing how we can do care plans using real data, and actually putting it into a state system where it will be used from that point forward.

The other thing that is coming are sort of our next generation of profiles. This is the EHDI workflow document. We are starting to move forward on this one. We don't want to just stop providing information, again it's this bidirectionality. We want to start looking at how can we get referral systems built into this. Where does information need to flow? Which providers need it? When do they need it? So this is basically the basic flow that we are working with. We're working with another committee at IHE called the patient care coordination. They're really looking at EHDI right now as sort of being out there in the front once again and that is that patient care plan, the overall care plan for a patient.

That's where we change from early care plan to this plan of care, that this becomes one element of a larger care plan of a patient.

I thought Dr. Botkin would be here. I've got several slides in here for Jeff. I want to talk about privacy and security because oftentimes people get a little bit worried about the information being out there electronically.

I want to just point out some things about electronic records I will start first of all with HIPAA. There are actually three portions of HIPAA, but two of those are the privacy rules. These are the rules that HIPAA has created for all types of medical records, paper based and electronic. But there's also the HIPAA security rule. And these are the rules and standards that go up and beyond those that are required for paper-based records. It talks about all of the administrative, physical and technical safeguards that have to be in place to secure electronic records. Also on top of this, HITECH also put more standards on top of HIPAA. That includes that if there's been a disclosure of information of more than 500, that information has to not only be told to the patient, but also that disclosure has to be released to HHS and to the media. So again, we've got even a higher standard here that we do for just straight paper-based records.

TMS also has their measures for privacy and security on top of these. There is actually a core measure if you're going to have a certified electronic health record. This is one of those meaningful use core measures that specifically talks about privacy and security. Part of their standard is you have to control who has access to this information. You have to authenticate the person that's going to have that information. The information has to be encrypted. Upon receipt you have to get the acknowledgment that the information has been presented to you in an accurate format and that no information has been altered along the transmission.

It requires an automatic logoff, so you can't just have a person sitting at their desk walk away and have somebody else come in to it. And then there's also required audit log for HIPAA. Excuse me, this is for CMS, but that would also meet HIPAA regulations. And it's actually going to record the date, time, the patient identification, who is the user, any time a record has been modified or even accessed, or if it's been disclosed, or if any information has been deleted. This is a requirement well above of what we're seeing anything for paper-based records.

I will just briefly talk about this is another committee at IHE. This is the information technology infrastructure group. These are the super geeks. These guys scare me. I can't understand -- every two to three words coming out of their mouth, I don't understand what they're saying. These guys are talking about what is under the hood. They've got dozens and dozens of profiles of how information moves. But one of those is the ATNA integration profile. And this basically is a profile for vendors that really talks about how do you establish, meet the security and policy procedures, about confidentiality. They've also created the basic patient privacy consent. So this is a way

that we can actually take information to record consent. So it's a way for us to look at privacy consent issues. There is a lot of people working on a lot of things.

People say, well, where do I start? There is a lot of places you can start getting involved, and I've just listed some of the major ones here. Pay attention to what is going on at the HIT policy committee, because this is where most of the big decisions that are getting recommended, this is a FACA committee reports to the HHS, to the Office of the National Coordinator and the CMS. This is where most of the gut work is working in terms of policy. They also have the standards committee. You can pay attention to that one. The standards interoperability initiatives, these are open to the public. There are lots and lots of these initiatives going on. I've spent several hours a week on many of these initiatives. You can get involved in HL7, specifically the committee we are working with is public health and emergency response or PHER. They hold meetings several times a year and these are the ones that are creating those standards, national standards. And then Integrating Healthcare Enterprise where we've been doing a lot of our EHDI work. Again, I give you the link here for all the profiles, and again just remind you that next week the profile is going up for public comment. We would love to have people review it.

I guess I finished earlier and I will be happy to take questions.

Joseph Bocchini: Thank you for the excellent presentation, it is remarkable to learn how complex it is to get some of this done, it is good information.

Let's open this for comments from the committee first, or questions.

If none from the committee, let's go to liaisons. Freddie?

Freddie Chen: Thanks. Freddie Chen from the AAFP. It is remarkable the amount of progress we've made from where we were just several years ago. But from your presentation I'm still trying to figure how close we are to me being able to look up screening results for a new child that comes to my clinic that was born somewhere else.

John Eichwald: I don't know how many electronic health vendors there are out there, but there is a lot. This is a moving target, and a lot of people talk about we are trying to fix the airplane midflight. And CMS keeps changing where we're going to land. And what are requirements for takeoff? It is certainly a moving target. I see some real progress. I think we're getting close to that tipping point where we're going to see it. Right now both hospitals and providers get incentives for using certified electronic health records to submit information to Medicare and Medicaid. In 2017 there is no longer an incentive. You actually get a reduced reimbursement. And so there is truly an incentive here for the vendors to be working with the public health hospitals and providers to make sure they are using a certified electronic health record.

Another thing that I did not put in here is that we are also looking at physician quality reporting initiative system to try to see if we can actually see if we can get physicians reimbursed for reporting to a registry. The question comes up, could EHDI be considered a childhood hearing loss registry. So the physician would report data to there, they can get an incentive. Are we there yet? No. We're seeing, like she said, we're seeing progress. The ball is rolling, and the ship has left the port, the train is out of the station, the horses are out of the barn. One of the things that sort of upsets me is that we need to be part of this discussion. Public health in particular has to be part of this discussion. And if we don't, we're going to be towed behind here and not the get the kind of information we need. I'm sorry. That was a long answer.

Michael Watson: I'm trying to figure out what NANI is. NANI? Is it an API that allows for exchange between particular information systems?

John Eichwald: I'm going to defer to probably the person who knows the most in the room and that's Terese Finitzo from OZ who helped write the profile for NANI.

Terese Finitzo: NANI is an HL7 message, so it is not an API. It is a true message and it is admission message. And so out of the electronic health record, comes this – it's an HL7 message and it provides basic demographic information on the baby. What we had to do for NANI, and this was really amazing, was when we were working with – when IHE was working on trying to get John's work with EHDI and Michelle Williamson's work with National Center for Health Statistics, we said to these guys, we need to know about the newborns. Can you send us a message on a baby who has been born in your hospital? And the answer was no, there is no newborn message. They could send us an admission message of everybody born. So Epic, GE, Cerner. So we literally had to say we don't want everybody. We just want you to constrain the admission message that you use to bill for a patient, to let people know that baby needs a test. We just want that on babies, and that's what NANI is. It's a baby message.

I want to say one more thing, and that is IHE is a great place for all of you, particularly the newborn blood spots screening or the critical congenital heart disease screening people. Because EHDI is where it is because of John, and he's going to probably be embarrassed that I say that, as well as because of Michelle Williamson, the National Center for Health Statistics.

So you've got a vital registration profile, and you've got all the EHDI profiles because he was at IHE to say, hey, I think this would be a good idea. So it is a great place for you guys to show up and push your own agenda. And that is what I think you want to do, push your agenda, whatever that is.

John Eichwald: And the other, it's version 2. Two five, two five one.

Michael Watson: I was just trying to get through to Freddie's interest in the results on the babies and whether they screen positive. It's nice to know they were screened. I'd

like to know if they were positive for something. The genetics part of EHR are an absolute mess. None of the companies make them. They make every group adapt it to their own needs, so I'm just trying to figure out how this communication takes place when you're having to brew your own.

Terese Finitzo: It gives you a pretty good standard. The admission message is about the simplest messages that hospitals can do. The problem still is, is they're pretty good communicating within their system, but they still have trouble communicating from inside the hospital out to you. But they just have in version 2.5.1, it's about the simplest message you can get. It's not going to give you everything on the blood spots card, but it could give you laboratory denominator that the baby, how many babies at St. Elsewhere Hospital.

John Eichwald: And EHDI could be used, as I say, as a basis. EHDI is pretty simple. I mean, we're not that deep in terms of what information we really have to capture. We want to know if the kid was screened. We need to know 1, 3, 6. We eventually want to get to outcomes. And so we've been able to move fairly quickly in EHDI. I mean, blood spot, we've got a whole bunch of issues here. Immunizations, we have to look at a long term. So I'm not saying that we've got it solved. But I think we've established a base here that other public health programs can build upon, but it's not a solution as of yet.

[Unidentified speaker]: I think NANI is very interesting. I'm curious about the timing of NANI because it would have to have occurred prior to sending us the blood spot card so that we can link it because without that, you can send me all the demographics but if I can't link it to the blood spots –

John Eichwald: The thing that's nice about NANI –

[Unidentified speaker]: And the hospitals haven't been able to do that yet.

John Eichwald: There are hospitals who are implementing NANI now. The thing that's nice about NANI is it's part of ADT, admission, discharge, and transfer. Any time a change is to that record, that information would be captured by public health. So if there's been a name change, that gets -

[Unidentified speaker]: I understand how it would work very nicely and I've seen pieces of it work very nicely for a week or two weeks after the birth. But when I'm working with blood spots that are supposed to be taken ideally around 24 hours of life and there's no medical record established for the baby and the baby is going underneath the mother's medical record number, then I need a physical card. Well, I need a physical card anyway, but I need some other way to link it. I need the timing to be very fast.

John Eichwald: Yes. That's why I think it needs to be electronic and have a patient identifier. Could be one of those things we're looking at so we can do some of that matching, and that's part of the NANI profile.

[End of recording]