The Advisory Committee on Heritable Disorders in Newborns and Children

HRSA Meeting

Washington, D.C.

August 02, 2018

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A P P E A R A N C E S

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JOAN SCOTT, Health Resources and Services Administration, Maternal and Child Health Bureau

DESIGNATED FEDERAL OFFICIAL:
CATHARINE RILEY, Ph.D., MPH, Health Resources and Services Administration, Genetic Services Branch, Maternal and Child Health Bureau

ORGANIZATIONAL REPRESENTATIVES:
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SIOBHAN DOLAN, M.D., MPH, March of Dimes, Department of Obstetrics & Gynecology and Women's Health, Albert Einstein College of Medicine and Montefiore Medical Center
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OTHERS:
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MARCI SONTAG, Ph.D., Director, NewSTEPs 360, Colorado School of Public Health, Director, Center for Public Health Innovation, CI International

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P R O C E E D I N G S

DR. JOSEPH BOCCHINI: Thank you,

Operator. Good morning, everyone. I would like to add my welcome to you. This is the third meeting of the Advisory Committee on Heritable Disorders in Newborns and Children for 2018. We will begin the meeting by taking a roll call.

So, going alphabetically: the Agency for Healthcare Research and Quality, Kamila Mistry? She may or may not be available this morning but will be on the call on and off during the -- during the day.

(No audible response)

DR. JOSEPH BOCCHINI: Mei Baker?

(No audible response)

DR. JOSEPH BOCCHINI: If you'll answer with "here."

(No audible response)

DR. JOSEPH BOCCHINI: Susan Berry?

(No audible response)

DR. JOSEPH BOCCHINI: So, if your phone is on -- on mute, please unmute it. We're not hearing any responses.

Jeff Brosco?

DR. JEFFREY P. BROSCO: I'm here.

DR. JOSEPH BOCCHINI: Thank you. Centers for Disease Control and Prevention, Scott Grosse will be here this morning. Scott?

DR. SCOTT GROSSE: I'm here.

DR. JOSEPH BOCCHINI: Food and Drug Administration, Kellie Kelm?

DR. KELLIE B. KELM: Here.

DR. JOSEPH BOCCHINI: Health Resources and Services Administration, Joan Scott?

MS. JOAN SCOTT: Here.

DR. JOSEPH BOCCHINI: Cynthia Powell?

(No audible response)

DR. JOSEPH BOCCHINI: The National Institute of Health, Melissa Parisi?

DR. MELISSA PARISI: Here.

DR. JOSEPH BOCCHINI: Annamarie Saarinen?

(No audible response)

DR. JOSEPH BOCCHINI: Scott Shone?

DR. SCOTT M. SHONE: Here.

DR. JOSEPH BOCCHINI: Beth Tarini?

DR. BETH TARINI: Here.

DR. JOSEPH BOCCHINI: And our DFO, Catharine Riley?

DR. CATHARINE RILEY: Here.

DR. JOSEPH BOCCHINI: So, for our organizational representatives, American Academy of Family Physicians, Robert Ostrander?

DR. ROBERT OSTRANDER: Here.

DR. JOSEPH BOCCHINI: American Academy of Pediatrics, Debra Freedenberg?

DR. DEBRA FREEDENBERG: Here.

DR. JOSEPH BOCCHINI: American College of Medical Genetics, Michael Watson?

DR. MICHAEL S. WATSON: Hello, I'm here.

DR. JOSEPH BOCCHINI: American College of Obstetricians and Gynecologists, Britton Rink?

(No audible response)

DR. JOSEPH BOCCHINI: Association of Maternal and Child Health Programs, Jed Miller?

DR. JED MILLER: Here.

DR. JOSEPH BOCCHINI: Association of Public Health Laboratories, Susan Tanksley?

DR. SUSAN M. TANKSLEY: Here.

DR. JOSEPH BOCCHINI: Association of State and Territorial Health Officials, Chris Kus?

DR. CHRIS KUS: Here.

DR. JOSEPH BOCCHINI: The Department of Defense, Adam Kanis, is unavailable for this meeting.

Genetic Alliance, Natasha Bonhomme?

MS. NATASHA F. BONHOMME: Here.

DR. JOSEPH BOCCHINI: March of Dimes, Siobhan Dolan?

DR. SIOBHAN DOLAN: Here.

DR. JOSEPH BOCCHINI: National Society of Genetic Counselors, Cate Walsh Vockley?

MS. CATE WALSH VOCKLEY: Here.

DR. JOSEPH BOCCHINI: Society for Inherited Metabolic Disorders, Shawn McCandless?

DR. SHAWN MCCANDLESS: Here.

DR. JOSEPH BOCCHINI: Thank you. Let's just go back and check for Mei Baker?

(No audible response)

DR. JOSEPH BOCCHINI: Susan Berry?

(No audible response)

DR. JOSEPH BOCCHINI: Annamarie Saarinen?

(No audible response)

DR. JOSEPH BOCCHINI: Okay.

DR. CATHARINE RILEY: Annamarie Saarinen just logged on to the webinar. And we -- we can see you on the webinar; are you on the phone, as well?

(No audible response)

DR. CATHARINE RILEY: Okay.

DR. JOSEPH BOCCHINI: All right, well, let's go forward, and -- and -- and -- and we'll catch these other members in -- in a few minutes.

Next on the agenda is a -- is approval of the minutes of the May meeting. The Committee received the draft minutes to review prior to the meeting. We've incorporated the revisions that were received as of this morning and sent the revisions out to the Committee.

In addition, we have received some additional edits. Each of these edits are -- are merely for clarifying technical information.

They're minor edits and correcting typos, and there is no real change to any of the substance of -- of the -- of the -- of the meeting minutes. So, we will go forward with the vote for approval of the May minutes.

So, we'll start with Mei Baker?

(No audible response)

DR. JOSEPH BOCCHINI: Susan Berry?

(No audible response)

DR. JOSEPH BOCCHINI: I approve.

Jeff Brosco?

DR. JEFFREY P. BROSCO: Approve.

DR. JOSEPH BOCCHINI: Scott Grosse?

DR. SCOTT GROSSE: Approved with the additional edits that Carla submitted.

DR. JOSEPH BOCCHINI: Could you repeat that? We didn't hear it well, Scott.

DR. SCOTT GROSSE: Okay. Approved with the additional edits that Carla Cuthbert submitted.

DR. JOSEPH BOCCHINI: Yes. Thank you.

Kellie Kelm?

DR. KELLIE B. KELM: Approved.

DR. JOSEPH BOCCHINI: Kamila Mistry?

(No audible response)

DR. JOSEPH BOCCHINI: Melissa Parisi?

DR. MELISSA PARISI: Approve.

DR. JOSEPH BOCCHINI: Cynthia Powell?

(No audible response)

DR. JOSEPH BOCCHINI: Annamarie Saarinen?

MS. ANNAMARIE SAARINEN: Approved.

DR. JOSEPH BOCCHINI: Joan Scott?

MS. JOAN SCOTT: Approved.

DR. JOSEPH BOCCHINI: Scott Shone?

DR. SCOTT M. SHONE: Approved.

DR. JOSEPH BOCCHINI: And Beth Tarini.

DR. BETH TARINI: Approved.

DR. JOSEPH BOCCHINI: Thank you, all.

Next item is a -- a new addition to the RUSP. As you know, in March, on behalf of the Committee, I sent a letter to the Secretary regarding our recommendation to expand the Recommended Uniform Screening Panel to include the addition of spinal muscular atrophy due to

homozygous deletion of exon 7 and SMN1.

We received a letter from the Secretary of Health and Human Services on July 02, 2018, that he had accepted the Committee's recommendation. In his response, the Secretary has asked that the Committee provide a report within 2 years, describing the status of implementing newborn screening for SMA, including clinical outcomes of early treatment and any potential harms for infants diagnosed with SMA.

The letter and its -- and the full SMA evidence review report are now available on the Committee's website.

I want to personally thank everyone who contributed to the nomination and to the review of the evidence of SMA. In particular, I want to thank the Committee members for their comprehensive review of the evidence and the discussion that resulted in the decision to move this on to the Secretary with our recommendation. So, thank you.

Next on the agenda is a call for

organizational representatives. Got the next slide? So, as you're all aware, the Committee really values the expertise and input from the organizational representatives. On the Committee website is an invitation for organizations interested in being considered for formal representation at Committee meetings. Criteria used by HRSA for selection of organizations are also listed on the website and summarized on this slide.

The Committee has received some applications through this mechanism. HRSA will now also soon put out a call for organizations that wish to be considered. I want to thank the organizations that have already expressed interest for their patience in this process. All applications received thus far will be considered, along with any additional organizations who wish to apply for serving as formal organizational representatives to the Committee.

Next slide. Also want to introduce a new

organizational representative for the Society of Inherited Metabolic Disorders is Dr. Shawn E. McCandless. Dr. McCandless is a Visiting Professor of Pediatrics and the Section Head for Genetics and Metabolism at the University of Colorado, Andrews School of Medicine, and Children's Hospital of Colorado.

Dr. McCandless is a graduate of Temple University's School of Medicine in Philadelphia, completed his pediatric residency at the University of Wisconsin at Madison, and clinical and biochemical genetics training at Case Western Reserve University. Dr. McCandless has worked as a general pediatrician in the Northern Navajo Medical Center in Shiprock, New Mexico, and held faculty positions in genetics and metabolism at the University of North Carolina, Chapel Hill, and at Case Western.

He previously served on the Ohio Department of Health Newborn Screening Advisory Council for 12 years. He is currently a Co-PI of the Urea Cycle Disorders Consortium of the NIH

Rare Diseases Clinical Research Networks. His research has focused on inborn errors of metabolism and the Prader-Willi syndrome. He is board certified in pediatrics, clinical genetics, and clinical biochemical genetics. Dr. McCandless is a fellow of the American Academy of Pediatrics and the American College of Medical Genetics and serves on the Board of Directors of SIMD.

So, I'd like to welcome Dr. McCandless as a organizational representative to the Committee and ask that all of us give a warm welcome to him. So, welcome.

Also want to --

DR. SHAWN MCCANDLESS: Thank you.

DR. JOSEPH BOCCHINI: Also want to thank Dr. Greene for her years of service as the representative for the Society of Inherited Metabolic Disorders and -- and all of her work and her involvement at Committee meetings and workgroups, and certainly look forward to Dr. McCandless continuing that tradition of providing

good service to the Committee. So, we appreciate your involvement and the involvement of your organization.

Next slide. As I mentioned at our last meeting, when implementing evidence-based decision-making, it's necessary to periodically evaluate the processes that are in place. And as I had mentioned at the last meeting, we made the decision to take a closer look at the condition-nominating process, including looking at the options for nominating a condition and removal of a condition from the RUSP.

As we go forward, we will also be assessing the entire condition review process. As you know, we talked about establishing a steering committee and -- and moving forward with the plan to complete this review. The evaluation will include how the evidence review is conducted, the components included, how the evidence is presented to the Committee, and how the decision matrix is being used. I'd like to see us update the evidence -- the decision

framework using the latest approaches for using evidence to successfully develop public health policy.

So, the Committee is working with HRSA to initiate this process, so please stay tuned. You will hear the plans that they have evolved relatively soon.

The next plan for the Committee is a -- an evaluation or an assessment of -- of the -- what has happened with the implementation of new conditions that have been added to the RUSP. So, again, over the next year, we also plan to take a look at the impact of adding the more recently approved conditions to the RUSP. We'll take a retrospective look on how the implementations have gone -- in particular, were the estimated time frames accurate? Were there barriers and challenges encountered that we did not anticipate and that states did not anticipate in implementing a new condition? Were there any unexpected challenges? We also want to take a closer look at the clinical and public health

implications of adding conditions with known delayed onset and severity. The Committee is also working with HRSA to initiate these efforts.

Next slide. The future meeting dates are on this slide. The next meeting will be held on November the 1st and 2nd, 2018. It will be an in-person meeting at HRSA headquarters in Rockville and available by webcast.

There has been a change in the dates of the spring 2019 meeting. It will be scheduled -- it was scheduled for May 9th and 10th. It will now be held April 22nd and 23rd. So, those of you who were planning for that meeting, please note this change.

It is -- it is expected that both of these meetings -- that this meeting in April will also be in person and available by webcast. And the meeting dates for -- through 2020 can be found on the Committee's website.

So, meeting topics for today: We have a presentation on risk assessment in newborn screening, followed by a presentation on

improving timeliness in newborn screening. We will have the workgroup updates and a report on long-term follow-up in newborn screening and a second report on technology in newborn screening that have been underway for a period of time.

Next slide. So, I'm going to turn this over to Catharine, now, for some business about the Committee. Catharine?

DR. CATHARINE RILEY: Great. Thank you, Dr. Bocchini. Good morning, everyone, and -- and welcome to those who have joined the webinar from the many different time zones across the U.S. and elsewhere. We know it's early for some of you. So, thank you for joining us.

I do have a few reminders and some logistics to go over this morning. This advisory committee's legislative authority is found in the Newborn Screening Saves Lives Reauthorization Act of 2014. This legislation established the Committee and provides the duties and scope of work for the Committee.

However, all Committee activities are

governed by the Federal Advisory Committee Act, which sets the standards for the establishment, utilization, and management of all federal advisory committees. As a committee member on a federal advisory committee, you are subject to the rules and regulations for a special government employee.

I also want to take this opportunity to remind the Committee members that as a committee, we are advisory to the Secretary of Health and Human Services, not to Congress. For anyone associated with the Committee or due to your membership on the Committee, if you receive inquiries about the Committee, please let Dr. Bocchini and I know prior to committing to doing an interview.

I also must remind Committee members that you must recuse yourself from participation in all particular matters likely to affect the financial interests of any organization with which you serve as an officer, director, trustee, or general partner, unless you are also an

employee of the organization or unless you have received a waiver from HHS authorizing you to participate. When a vote is scheduled or an activity is proposed and you have a question about a potential conflict of interest, please let me know as soon as possible.

So, according to the Federal Advisory Committee Act, all committee meetings are open to the public. If the public wish to participate in the discussion, the procedures for doing so are published in the Federal Register and are announced at the meeting.

For this meeting, in the Federal Register, we said there would be a public comment period, and for this meeting, we did not receive any requests to make an oral comment, and we also did not receive any written comments ahead of time. There is a brief section after lunch, and if there's anyone interested, please -- please let us know by -- by raising your hand, and if we have time, we'll be able to get to that. But, again, we did not receive any requests ahead of

time.

So, public participation should be advised that the Committee members are given copies of all written statements submitted to them ahead of time, and in this case, they didn't receive any as none were submitted. Any further public participation will be solely at the discretion of the Chair and the DFO.

So, before I move on, any questions from the Committee?

(No audible response)

DR. CATHARINE RILEY: Okay. So, for the webinar, since this is a -- Oh, sorry, before we -- before we go on, I see -- Dr. Berry, did you have a question?

(No audible response)

DR. CATHARINE RILEY: Okay. We'll move on, but I think Dr. Berry had a question about logging in. So, while we're holding that, we'll move on.

So, for Committee members, organizational reps, and speakers, please use the "raise hand"

function if you have a comment or a question. So, this is -- at the top of your screen, you can see the figure, and it has a little hand. You can click on that to raise your hand. We'll note that in the queue. So, once we've noted that, the Chair, Dr. Bocchini, will call on you in order of your request. We please ask that you unmute your phone lines before providing comment or question. Also, please remember to state your first and last names every time you provide a comment or question just so we can ensure proper recording for the Committee transcript and minutes.

For all of the meeting attendees that are watching the webinar and listening in, thank you, again, for joining. We cannot receive audio through the webinar, so you'll be able to hear the webinar, but we cannot receive audio back. So, there will be no functionality as far as raising hands, et cetera.

Okay. So, if -- I'm going to open up one last time. If there's any questions from

Committee members, organizational reps, or speakers that have an open line --

(No audible response)

DR. CATHARINE RILEY: Okay.

DR. JEFFREY P. BROSCO: Hi, this is Jeff. I'm pressing the button. Are you not seeing it?

DR. CATHARINE RILEY: I am seeing -- Yes. So, Dr. Brosco, go ahead.

DR. JEFFREY P. BROSCO: Sorry, I just had a request from the public to be able to participate in -- in the webinar, and I wonder if we could send around a link for folks who want to participate. They apparently tried to register but were unable.

DR. CATHARINE RILEY: They tried -- what, they tried to register this morning, Dr. -- This is Catharine Riley. Were they trying to register today?

DR. JEFFREY P. BROSCO: Yes.

DR. CATHARINE RILEY: Okay. If they could send out the link -- Yeah. So, Dr. Brosco, if you could -- if there are folks you

know are interested, if you can have them email Alaina Harris?

DR. JEFFREY P. BROSCO: Alaina Harris, okay. Thank you.

DR. CATHARINE RILEY: Yes. And we will get them connected.

DR. JEFFREY P. BROSCO: Thank you.

DR. CATHARINE RILEY: Okay. So, we have -- I have Dr. Berry. Did you have a question?

(No audible response)

DR. CATHARINE RILEY: Okay. And last one is Dr. Ostrander. Did you have a question? You-

DR. ROBERT OSTRANDER: I just, early on, wanted to quickly mention -- this is Bob Ostrander, Academy of Family Physicians -- just to get people's ears perked up about our comments from Follow-Up and Treatment, later on, based on the Secretary's response to our addition of adding SMA to the RUSP and the request for early follow-up information on that condition. I think that our comments from Follow-Up and Treatment, later, about some thoughts about trying to

tighten that process up are going to be germane, and I just wanted to have people make that link in their mind going forward.

DR. CATHARINE RILEY: Okay, great. Thank you, Dr. Ostrander.

So, before I proceed, just a reminder: There -- there is no functionality with the webinar to communicate with, you know, myself or -- or Dr. Bocchini. So, you can click the "hand-raise" function. Other than that, we will have to utilize the open conference line for communication. So, I just wanted to offer that reminder. Or you can email -- this is Catharine Riley. You can email me or Alaina Harris if you have questions or issues with the webinar as -- as we proceed.

Okay, if no further questions -- I don't see any other hands raised by Committee members or registered speakers -- I'm going to turn it back over to Dr. Bocchini.

DR. JOSEPH BOCCHINI: Thank you, Catharine. We're ready to have the first

presentation. This is on Risk Assessment in Newborn Screening. As you know, this has been an issue that the Committee has been considering for a period of time, and we've had a number of presentations from stakeholders, and our Laboratory Standards and Procedures Workgroup have been involved with APHL in their work on developing a -- a guideline with resources for the states. And so, this morning, we will have a presentation from Dr. Kelm concerning where we are with this issue.

Dr. Kelm is a Committee member, Chair of the Laboratory Standards and Procedures Workgroup. She is going to provide this update, and after this update, we will have time for the Committee and -- and others to discuss possible next steps or ideas for activities that the Committee might pursue in this topic area.

So, Kellie?

DR. KELLIE B. KELM: Yes.

DR. JOSEPH BOCCHINI: Are you ready to go?

DR. KELLIE B. KELM: I'm ready.

DR. JOSEPH BOCCHINI: All right, go ahead. The floor is yours.

DR. KELLIE B. KELM: Excellent. So, next slide, please. So, most of what I'm going to be talking today is an update on newborn screening risk assessment in the context of the discussions that this committee has been having for more than the past year.

And we had a workgroup meeting on Monday, July 30th, and we discussed many things, including the APHL risk assessment guidance documents that the Committee has heard about in the past, an update from the CDC on their progress with harmonizing newborn screening assays. We heard some information from APHL on the NewSTEPs data repository, as well as some of the other things they've been doing to help laboratories during their technical assistance site visits and risk assessments. And, lastly, we talked about some future direction. So, that's what I'm going to summarize for you all

today.

Next slide. Next slide, please.

DR. CATHARINE RILEY: Hi, Kellie, this is Catharine Riley. We did advance the next slide. Are you seeing it on your end on the webinar?

DR. KELLIE B. KELM: Nope, I'm not. But I can bring it up on my own computer and try to keep along, but I'm still on the agenda slide.

DR. CATHARINE RILEY: Okay. So, we're going to go from the agenda slide to the Committee discussion on cutoffs slide.

DR. KELLIE B. KELM: Yeah.

DR. CATHARINE RILEY: Okay, yep.

DR. KELLIE B. KELM: So, just a reminder -- just a reminder that there've been several presentations to the Committee on newborn screening and the process for setting cutoffs and some of the challenges and limitations in -- that laboratories experience, including biological, analytical variability, et cetera. And so, we had a series of -- of presentations in 2017, and APHL started working on this risk assessment

guidance document, and the outline of that document was presented to our workgroup in August of 2017.

Next slide. So, after giving some very high-level feedback in August to the outline, we received a draft document for review, and the Workgroup provided feedback in November. And the draft document was widely provided to the newborn screening community in January, and the Workgroup noticed that many of -- most of the suggestions have been addressed in the draft. And the Workgroup provided additional clarifications to the first author, and that's Joe Orsini, who provided that on to the -- the authors working on the document.

Next slide. So, just a reminder of what had been discussed in February, sort of the conclusion of the Workgroup, and what we had also communicated to the Committee in our review of the document: The document does describe the scientific process behind establishing and validating cutoffs. The document will be

valuable to state newborn screening programs, a very good resource. APHL intends for this to be a living document and will revise the document over time. It does not include best practices for screening for all conditions, and it does not harmonize newborn screening tests across states.

Next slide. So, just to recap what -- After a presentation of the current draft by Joe Orsini to the Committee at our February meeting, the Committee decided a vote was not required on the document, acknowledged the document's value, and recommended that APHL continue to refine and improve it. It was also agreed that the Lab Standards and Procedures Workgroup should focus on what could be done to address public access issues and better ways to collect and store data on false positive results. And so, that was, sort of, the last update that we had.

So, about a week and a half ago, that final document was provided to the Workgroup, as well as -- as provided to the Committee, and at our July 30th meeting, Workgroup meeting, we

heard a brief presentation from the APHL on the changes to the document and had more discussion on -- on the document.

Next slide. So, per feedback from the Advisory Committee, a summary table was added and highlights -- generally, the -- the -- the table just takes highlights from the text and puts them in a table that is easier to look at the different types of methods for -- in terms of what type of cutoffs -- fixed, floating, multiples of the median, et cetera -- and their -- what they're generally used for and some functionalities and considerations.

The QA/QC Subcommittee Workgroup also -- out of APHL -- had consulted with experts in the field on the different methodologies, had updated these documents to reflect more accurate information on these methodologies. Also, there were changes to the document to, I would say, update the -- the tone, and people felt that it also -- the edits provided a better balance in terms of discussing all the different methods and

available technologies in the document.

Next slide. So, the current status of the document is that it's very close to a final draft. Recently, the -- the group had resolved several comments that they'd received asking for additional clarifications, mainly from Mayo, in terms of the section on CLIR. We -- they did receive additional, recent comments from Stan Berwick on some errors on the section of -- on the -- the multiples of the medians, and so that section will be updated. So, it's not quite final yet, but the -- it's -- it's very close to being final once those -- those comments are addressed.

So, as I said on a previous slide, this is a living document and will have changes made, updates, as more information is made available. And at this time, APHL plans to post it on their website in the very near future, they're hoping within the next couple of weeks.

So, that's the update on the document, and I have a question for the Committee on -- on

the document at the end of my talk.

And so, we'll move on to -- next slide -- what we heard in terms of an update from CDC. And so, this is, as we've heard before, part of their Quality Assurance group and harmonization activities, and so Kostas Petritis talked about normalization of the MS/MS biomarker results when he provided this update to the group.

Next slide. So, if you recall from his previous presentation and what we've heard, also, in this discussion about cutoffs is, mass spec biomarker measurements and -- and cutoffs can vary significantly among different labs. And just a reminder that over 70% of the disorders on the RUSP can be screened by mass spec. So, the -- the -- the different analyte results in cutoff values varies, and the major contributors are different extraction methodologies. You have labs using derivatized versus non-derivatized methods. Few labs account for analyte recovery; most labs don't. And then, there's the use of additional or different analytes per disorder or

second-tier screening and, of course, other factors in populations, instrumentation, internal standards, and calibration techniques.

Next slide. So, Kostas actually showed us, in the last -- in his presentation, how CDC is performing this harmonization by getting state labs' results with their QC materials, and then they're using the proficiency testing results to validate that their normalization process is working. And so, this is just one of the slides that Dr. Petritis showed us in terms of the different results, and then using that to harmonize the results.

Next slide. He also showed the before and after in terms of the results that we're getting and how normalization improved the difference amongst the labs when -- when they had performed the normalization and showed that, for example, an outlier, as shown in the circled dot in the right side, could be pointed out to the labs so they could reevaluate their cutoffs, even -- because normalization would, obviously, really

expose the -- the potential outlier in terms of cutoff here.

So, next slide. So, we got a short presentation on what CDC has been up to in the last few months since their presentation on their normalization project.

Next slide. So, currently, CDC is building a web interface to visualize the normalization results, and this would -- would -- would be available to the laboratories for them to be able to do this comparison for themselves. And so, this is currently in design phase, but they were showing us what they intend to be able to present to the laboratories for them to come in and look at how they compare to other laboratories.

Next slide. In addition, they were providing us updates that, obviously, they cannot do normalization for analytes that aren't included in their QC materials. So, they are working to add more analytes to their QC materials. And so, their current production,

which will be shipping early next year, will contain the additional analytes listed in the list below. And CDC is considering adding additional analytes, including ASA and those other ones in the list down at the bottom, in order to allow them to -- to normalize these results in future -- in -- in -- in future time periods.

Next slide. So, they did discuss some of the limitations of this current process. So, using proficiency testing samples to confirm that normalization worked has provided proof of concept but is not a long-term solution. This is only one measurement. Not all analytes are enriched in every proficiency testing event, and some analytes are outside in the -- in the -- proficiency testing samples are outside the dynamic range of the QC materials.

And there was also some discussion by our workgroup members that they would really like to see clinical samples used in order -- because of the different -- and limitations of PT samples.

And so, there was additional -- CDC's considering the creation of an additional QC specimen, with all analytes set at CDC cutoff, to be used for normalization and validation.

And so, that's the update we got on what CDC has been doing in this process for the last few months since their first presentation.

So, next slide. So, we received an update from Jelili from APHL on some of the current work that NewSTEPs has been doing, helping with quality improvement, that also helps with cutoffs and risk assessment and also talking about future activities for NewSTEPs.

Next slide. So, just a reminder that NewSTEPs has -- has several goals, and so I -- I wanted to include them all here and -- and -- while we get to the one that helps with risk assessment cutoffs, but just a reminder that their first goal is communication and outreach, and they have a lot of activities, obviously, to -- in -- in -- in that space.

Next slide. Goal two is their role with

continuous quality improvement and data-driven outcome assessments in newborn screening by providing centralized data repository, their dynamic data infographics and visualization tools, and supporting integration of HIT frameworks, including HL7 messaging.

Next slide. And, lastly, technical assistance -- They have a technical assistance resource center that proactively provides training, addresses challenges, and supports program improvement, and a -- a large part of this is their comprehensive site reviews, as well as focus site reviews that they've been forming, where they send groups of experts to laboratories that have requested the visits. And we know that in several instances, sites have actually requested help in -- from these groups in, sort of, reviewing their process for establishing and reviewing cutoffs and giving them feedback and helping them work on their -- their SOPs and improving them and -- and having these experts in laboratory -- lab procedures and follow-up has

been very helpful to laboratories in improving these processes.

Next slide. So, NewSTEPs, which recently awarded funding from HRSA, I believe, that will be starting next month, and you notice that as part of the quality improvement projects, there are five focus areas for the funding, and number two in that list is identification and follow-up on out-of-range results. And so, this is quality improvement, specifically in the area of cutoffs, establishing cutoffs, validation, risk assessment, and assessing how well that's working in terms of follow-up and providing information.

And, also, you know, we have had these discussions lately about improving assessments and collection of information on false negatives, and what that may also mean for systems. So -- so, this -- this -- these activities will probably be very helpful for laboratories as NewSTEPs rolls out these activities.

Next slide. And they also performed other activities, in this case, that CDC funded,

as we've been talking a lot about the QA/QC Subcommittee and their work on the risk assessment document. They have other activities, including education -- educational webinars, providing information and help to labs as new conditions are added to the RUSP, and other QA/QC activities and -- and -- and providing that -- that help. And we've heard before -- previously about the MBS Molecular Assessment program, and this program provides site reviews addressing molecular capability -- capacities, excuse me, in newborn screening programs, and there have been 22 program visits since January of 2011.

So -- so, that's -- I -- I think we look forward to hearing more from APHL and NewSTEPs about how the activities in this area are helping laboratories as they work on, you know, adding new conditions and -- and also, you know, working on SOPs, which the Committee recommends that -- that laboratories have in terms of how they are establishing their cutoffs and validating their cutoffs in a robust manner and revisiting those,

as well.

So, next slide. So, as we've been having these discussions about, you know, the attention that's been given to cutoffs the last, you know, a year and a half to two years, one thing that has come up in our discussions with the Committee, as well, and -- and -- and members of the Committee, which is the possibility of a cross-workgroup effort involving people from the Lab Standards Workgroup, as well as the Education and Training Workgroups. And so, what has -- what has emerged is whether or not there's ways to communicate the strengths and limitations of newborn screening to different audiences, because that seems to be a piece that is still missing as we have talked about the attention given to the issues with cutoffs in the media.

So, one of the ideas was to potentially create some sort of a tool or product to educate physicians, parents, and/or the public on newborn screening, but some of the questions, since we're just sort of talking about this possibility, is,

what would this look like, and who would be the target. And so, this is something that we've just, really, sort of, scratched the surface on.

Next slide. What -- in our discussions, what this has -- what we've envisioned in terms of a general message for any -- any of those target groups would be: describing what is screening, what is newborn screening, how newborn screening is different from other types of screening, and what are we trying to find. And what other -- the other thing that has come out of our discussions is the idea -- in terms of our messaging for physicians, is adding more information on the limitations of newborn screening and reminding them to act on the clinical signs and symptoms regardless of the newborn screening result, you know, not to assume that a negative is always a negative.

So, this is something that has come up that, you know, we've also briefly talked to some members of the Education and Training Workgroup on, but we would love to get some feedback from

the Committee on whether or not they think this kind of product or -- would be something that would be of interest to the Committee and of interest to the general population but then, as we said, you know, physicians, parents, public, very, you know, different target groups in terms of knowledge, interest, experience, and training. And we'd love to hear more from the Committee and others on what they think about something like this and whether or not it would help communicate to those outside of newborn screening about all of these issues that we've been talking about for the last two years.

So, my next slide is the last slide. Our -- so, our workgroup, we sort of ended with two questions that we wanted to put in front of the Committee for discussion at this point. So, you know, we have -- and -- and the Committee has received pretty close to a final document on APHL's risk assessment guidance document, you know, provided to -- just a reminder of what we -- the Committee had concluded in February in

terms of the document, but we did want to ask whether or not the Committee felt there was any change in their recommendations in terms of what to do with the document and whether or not there was anything the Committee wanted to do with the document.

And number two, the -- the question about whether or not there was any interest by the Committee in having a Lab Standards-Education Training cross-workgroup, sort of pursuing together creating this educational product in newborn screening for physicians, parents, and/or the public. And so, that was our -- our second question that we wanted to put in front of the Committee for discussion.

So, that's it for me. I'd love to hear some feedback from the Committee. Thank you very much.

DR. JOSEPH BOCCHINI: Kellie, thank you. That was a great presentation, and I think it sets the stage for a good discussion from the -- from the Committee and the organizational

representatives.

So, let's open the discussion. Again, use the "hands up" to indicate that you want to ask a question or make a comment, and then we'll -- we'll go through the list and move forward.

I think the questions that Kellie has raised, I think, are really excellent, and I want to frame them, also, in what we've spoken about before as to what other things can the Committee do or consider doing to try and -- and help this APHL document get the -- the use that it -- that -- that it needs, as well as help states in any way to make risk assessment work better. So, let's open that to discussion.

So, first question, comment, we have Dr. Berry. So --

(No audible response)

DR. JOSEPH BOCCHINI: So, Sue, we can't hear you. Is your line muted?

FEMALE SPEAKER: And we do not have Dr. Berry dialed in to the speaker line with an open line. If -- Dr. Berry, if you are online, could

you press "star 1," please?

(No audible response)

FEMALE SPEAKER: And I am not getting a response at all.

DR. JOSEPH BOCCHINI: So, if Sue is coming online, while we're waiting for that to happen, let's go to Mei Baker.

DR. MEI BAKER: Hello, can you hear me?

DR. JOSEPH BOCCHINI: Yes, we can, Mei.

DR. MEI BAKER: Great. I -- the -- in terms of any changes from February 2018, the recommendation -- and it just right now popped to my mind. I would like adding on one thing. I think it's wonderful CDC, going forward, will make QC material for laboratory do the validation.

Also, I think, is it possible, when CDC obtained QC data from each lab, ask them that on the report absolute numbers, can I -- can we also ask for multiple of the median? Because this one, you naturally normalize each set of data, then do the comparison knowing where they are.

And also, when return this data back to CDC, also provide their multiple of the median, their cutoff.

So, this way, this -- I think this the more efficient way to obtain the data, and CDC can analyze them in the deidentified fashion. And we all know our laboratory code. Then, we receive the feedback and allowed us to see where, you know -- each lab, where we are. Is something durable? So, this is a -- I think it's a part of the process that we can harmonize data, and I feel multiple of the mean, maybe, is the -- the -- the way we do that.

And second part I want to make some comments -- I think Sue, actually, can make a better comment. My understanding is, the Midwest Genetics Group is underway develop a set CME education data for primary care physician, and I think all the effort should be collaborate together in standard, you know, reinvent the wheel.

DR. JOSEPH BOCCHINI: Thank you, Mei.

Sue, are you on? Sue Berry?

DR. SUSAN A. BERRY: Yes, thank you. I -- sorry, I misunderstood about the issue on the webinar. My apologies.

DR. JOSEPH BOCCHINI: No problem. Go right ahead.

DR. SUSAN A. BERRY: I wanted to make the Committee aware of a project that's being undertaken in the Midwest Genetics Network, part of the HRSA-funded genetics collaborative. We have just had word from the American Board of Pediatrics that our MOC4 educational activity for physicians regarding education around newborn screening is -- has been approved.

The -- there are basically three training modules to this, and the three training modules are: an introduction to newborn screening and what it does and what it doesn't do; a -- and a module specifically on sharing results that are negative and what that means for families and how to share that information; and then the third module is on giving back either so-called

borderline or positive results. And the idea, I think, really harmonizes with what's being discussed for this educational product that -- that Kellie mentioned. We identified this based on a -- sort of a -- so the -- the Education Committee has a -- a broad spectrum of stakeholders, included -- including families, and they identified better education for physicians in newborn screening as -- as an important priority to them.

Obviously, they're a specialized group of folks, most of the time, when we -- when we have consumers in these genetics networks, but I -- I think the -- the upshot of it is that -- that the use of -- of educational activities is really important. People -- the -- the -- we -- we found, in studies, that where we've surveyed physicians in our own area that despite, I would say, relatively medically sophisticated understanding that they typically didn't understand the import of a negative screen and that that was something that really needed some

highlighting.

DR. JOSEPH BOCCHINI: Great. Sue, just a follow-up question for you: For the MOC, you need to be a member of the American Board of Pediatrics to --

DR. SUSAN A. BERRY: Yeah, unfortunately our --

DR. JOSEPH BOCCHINI: -- take the MOC?

DR. SUSAN A. BERRY: -- our first pass on it to get it out there, because we wanted to -- we wanted to get it done. So, for the first round of MOC4, we got it to APP because they could get it through the process most quickly, and we wanted to do that.

Our plan with this set of educational modules is to work with an additional provider to make those modules more generally available to providers at other levels of training, so that, for example, you could use it for PAs or as -- we -- we're hoping to be able to connect with the American Board of Family Medicine, for example, the -- we -- so that this could be more generally

available. We -- we wanted to make it available as soon as possible to our pediatric colleagues in our region, so that's where we started.

DR. JOSEPH BOCCHINI: That's great, and then, perhaps, maybe even through the -- for residency training-program modules, as well.

DR. SUSAN A. BERRY: Mm-hmm.

DR. JOSEPH BOCCHINI: So, thank you --

DR. SUSAN A. BERRY: Yeah, we'll --

DR. JOSEPH BOCCHINI: -- that's great.

So, next two Committee members are Scott Shone and then he'll be followed by Beth Tarini.

So, Scott?

DR. SCOTT M. SHONE: Good morning. So, a -- a couple things, and, Kellie, forgive me if I'm not tracking with the entire presentation. I don't know if that's just because of the webinar or whatever, but it seems to me that -- that this discussion, at least this morning, was a multi -- multi-faceted summary of some -- of -- of different activities, and I just want to be sure I'm clear on that.

And so, the first is, the APHL document, which we've been taking about -- which I think the final revisions have made it a lot stronger, but that's around the discussion of current state of cutoffs and -- and -- and how programs do that. But then Kostas is sort of painting a picture for the vision of where states could go -- and programs, rather, could go with taking this beyond just a cutoff discussion and maybe even helping to harmonize analyses across -- across the system with -- and -- and, again, please let me know if I'm misinterpreting this -- with the goal of, you know, sort of trying to get a -- a case where if a baby's screened in one program or another program, the interpretation is the same.

And the third is education of the appropriate people within the system on this topic and the topics associated with risk assessment.

So, I want to stop and make sure I have that correct, because I might have comments about that.

DR. KELLIE B. KELM: So, you -- you're correct, and I think that -- I think I should have done a better job introducing that I think a lot of the negative attention that was given to the systems in terms of screening -- what we heard was, you know, I guess several things: 1) you know, criticisms that the cutoffs weren't appropriate and that they were missing babies, 2) you know, the -- a lot of it was the fact that different programs had different cutoffs and different results. And I don't think -- while we often understand why that is, there, obviously, is also some interest in whether or not we can figure out some way to -- to normalize those results across states so that it makes more -- you know, it seems like the same results in Alabama as what you're getting in Missouri, whether or not that's, you know, going to happen or not.

And then, 3) you know, is there a way for us to provide some education, because a lot of the information that was in some of those

articles was incorrect or, you know, there is -- unfortunately, is some lack of knowledge about newborn screening and the limitations, I mean, getting that out in order to also address some of the incorrect information but also just making sure that we're providing the right information out there.

So, I think this is just addressing several of the different topics that came up in some of the media attention for newborn screening that happened a couple of years ago. So --

DR. SCOTT M. SHONE: Right.

DR. KELLIE B. KELM: -- that's how I tackled -- you know, we're -- we're sort of talking about activities to address each of those.

DR. SCOTT M. SHONE: All right. So, I -- to -- to the last point and, sort of, in terms of the -- the education and training -- I mean, we've had, at least since I've been a Committee member and -- and beyond -- and before that, there's been some great presentations by the

Education and Training Workgroup, you know, Beth and Cathy. Amy Gaviglio's given many presentations, and I -- So, I think that these topics have been addressed -- Genetic Alliance has been involved -- around, you know, these topics of information and who -- and who they need to go to.

And so, I don't think -- I'm not sure -- and -- and, again, I might be reading this wrong -- I'm not sure if I'm -- I'm understanding the goal, but I think that -- that some of this is already in progress, and I don't know that the Lab Standards Workgroup is going to contribute more to education than the Education and Training Workgroup. And, perhaps, the idea is more of assisting with dissemination of work -- of some excellent work that's already -- already been done or in progress. But -- but -- so -- so, I want to be cognizant of the amount of work that's already been done on that -- on the education topic by the other workgroups.

But to the point of risk assessment, I

mean, I -- I think that, you know, media attention obviously brought -- you know, brought the topic up, but the idea that there's widespread misclassification of risk in the newborn screening system -- I -- I don't think anybody has ever said that. And so, some of these efforts have been -- really made it clarifying why programs do what they do and -- and -- and, also, how they do what they do. But I want us to all be aware of that, you know, the work that's being done at CDC, the work that's being done to develop other post-analytic tools, are helping to -- are -- are -- are meant to help hone and -- and refine that risk assessment, you know, not only to avoid missed babies but to not -- to not overcall positives.

And so, I think, from a technical assistance standpoint -- and -- and you -- you mentioned NewSTEPs -- there's an opportunity there to -- to not only educate on why and what we currently do but build on what's out there to go towards the future. And so, I don't want to

get that lost in -- in this discussion around -- that I think that's --ultimately, the goal is improving risk assessment and -- and not suggesting that -- that -- that any of these activities are -- are sufficient in terms of just, this is how we do and why we do but rather that the system is always trying to improve and -- and make sure that the right babies are identified. And I'm not sure I'm articulating myself clearly enough, though.

DR. JOSEPH BOCCHINI: All right, thank you. Beth, and then that'll be followed by Jeff Brosco.

DR. BETH TARINI: So, my -- my questions actually follow from Scott's, and the first is to ask, what does the Lab Standards Group think that is missing from the current strategy of education? And what follows to answer that question is, do we have a understanding of, what is the full landscape of what we are doing now.

So, it sounds like -- and -- that APHL, the Committee on Genetics and Public Health, is

working on this. Genetic Alliance is working on these issues. So, I'm now wondering if the first step is actually just to assess the landscape of what we're actually doing, and then identify where there are either gaps which we're not covering -- either content or audience -- or whether -- and is it not an effective strategy. So, that's my first point. And then my -- and -- and question.

And then, my second is, the -- the -- the concerns about newborn screening and its portrayal in the media -- and I -- and I want for us to try to clarify, is this a perception issue, or is this an education issue, because there -- the distinction is important. If we are trying to change perception from the public, then I think our strategies may be different than if we are trying to change behaviors of physicians and parents.

And I'm not saying which one we should be doing, but -- but I think -- For instance, I would argue that we have -- we, often, are in an

echo chamber, and I don't think a lot of our information is getting out into the lay public. And if that is the problem, no matter how many education tools we create, they won't get to the lay public. I mean, then this issue of getting into the media, having conversations with reporters -- which has come up in our workgroup, and -- and Catherine is, I know, looking into this -- I -- I think that has to be considered as part of a strategy. Otherwise, I don't know that we've pierced the veil into the public with -- with the current educational strategy we have.

DR. KELLIE B. KELM: Yeah, this is -- this is Kellie, and I think -- I -- I think your first point is especially useful, that in some ways, what we almost need to do is understand the landscape. And I do think that, for example, also finding out more information about activities that are out there -- for example, what Dr. Berry talked about -- are -- are things that we would probably, you know, really need to assess before we ever build into, you know,

thinking that there was some need for some other product, but, you know, your second point obviously is -- is extremely valid, as well.

DR. JOSEPH BOCCHINI: All right, next is Jeff and then --

DR. JEFFREY P. BROSCO: Yeah, Jeff Brosco.

DR. JOSEPH BOCCHINI: -- followed by Mei Baker.

DR. JEFFREY P. BROSCO: So, I -- I -- I agree with and just wanted to emphasize what Scott had said, and -- and some of what Beth said, as well, that we -- we do want to be careful not to start making new policy or making big, new initiatives for what might be a -- a -- a small problem in terms of the number of -- of children who are missed. So, I think that's a really -- a good point that Scott made we've been trying to make all along.

And as Beth was pointing out, there is a huge amount going on already, and, you know, of course, Genetic Alliance and Baby's First Test,

there's a lot of trying to help people understand. And my one suggestion is that we want to think as much as we can about just-in-time information, and -- and that is that the -- the information education happens at the moment that either the -- that the person actually needs it.

And one opportunity for that, I think -- and my colleagues in Florida may -- may scream when they hear this, but when the test results get sent out to -- to the pediatrician or to the hospital, it seems to me that every state probably does something a little bit different on how much information they provide. And it might be that a good explanation of what those results mean and don't mean at that moment -- So, if, every time a pediatrician or a family doc got the newborn screening results, there was a clear statement about, you know, what this means, what the chances of false positives and false negatives are, that might be a way to do ongoing education at the moment when a provider needs it

the most. And that's just one example of trying to think about education in terms of just in time, at the -- the moment someone needs it. Thanks.

DR. JOSEPH BOCCHINI: All right, next is Mei Baker, followed by Melissa Parisi.

DR. MEI BAKER: I just want to -- after Sue's comments, I want to emphasize: I -- I -- I feel strongly is, indeed, we need general population education, but I think we perhaps shouldn't expect all the parents to understand all the nuance of the scientifical things behind the whole cutoff will come about, and I do feel a primary care physician is a very, very important group.

And to go back to Jeff was saying, in-time information: that's because most of the newborn screening laboratory or program, we do not have a direct relationship with the parents. And our primary care physicians are the people. And the -- in -- in terms of trying to figure out is address the perceive or behave or education, I

feel, due to the -- the foundation base is important is why I feel the work through the letter is so important, because this is continued-education based, and, also, other profession organizations, which is the target, get involved.

Another thing I feel: If we have this module available become part of the package of continue education, so it's -- it's ongoing. It's not a one-time effort. So, I feel, very strongly, emphasize for our primary care physician is so important because they're our window or our channel to the family. And, also, usually, primary care physician have relationships with family. What they say, family likely has the trust, has the belief. So, how they are relating information to the family is terribly important. If they have a fully good understanding what the newborn screening mean -- what's the pros, what's the cons, what's the limitation -- so they can relay to family in a more accurate way --

So, I -- I -- I -- I really think -- I think if the material the Committee members want to have a chance to review -- and that's good. I just feel -- agree with Scott, with Beth, was that we -- we came in and say we do activities and -- but we need to assess what the outcome, too.

DR. JOSEPH BOCCHINI: Thank you, Mei.

Melissa Parisi, and then Sue Berry back again.

DR. MELISSA PARISI: Hi, this is Melissa Parisi. I just wanted to make a comment about the public-education efforts, and this sort of stems from some of the discussions at an NBSTRN meeting that was just held yesterday and the day before. And -- and, really, keeping in mind the value of reinforcing the importance of retention of dried blood spots for public-health-related purposes, for research purposes, and for, you know, just in general, the value of these spots as a resource for the future.

So, I know we obviously want to ensure

that the public is educated about newborn screening, what it does and what it doesn't do -- and -- and I think, by extension, the public -- I'm also including physicians and caregivers, who, you know, are going to be encountering these incidents, but I also think we want to not forget about some of the additional value of this whole program and the value of the dried blood spots for future purposes.

I mean, we know that there has been a very negative campaign waged against retention of dried blood spots, and, you know, the tragedies that have occurred in Texas and Minnesota with regard to destruction of all those spots -- you know, we can't forget about that. And whatever types of educational efforts, I would like to see that they at least include some acknowledgement of the value of this -- of this really precious resource.

DR. JOSEPH BOCCHINI: Thank you. Sue Berry, and then Beth Tarini.

DR. SUSAN A. BERRY: This is Sue Berry.

As the materials are made available for a -- for the MOC4 use, I'm certain that the Education Committee and the Midwest Genetics Network would be happy to share that information for comment and -- and -- and just so you can see what we're doing and to see if it's of more general utility for further use. So, we -- the -- the obvious purpose is to make that a generalizable resource.

DR. JOSEPH BOCCHINI: Thank you. Beth?

DR. BETH TARINI: So, I have a proposal for us to consider, which is based on the fact of Jeff's point about one -- one-time -- or just-in-time information and Mei's point about the primary care providers. And the fact is, as we all know, these are rare occurrences, I would say, generally, in a physician's career. So, educating them -- I -- I think baseline education is important, but I think it will not overcome the fact that the education muscle is not going to be used frequently, because these physicians just will not see enough of these in their career to, sort of, be superb in their retention of the

knowledge base.

So, back to, then, what the states can do and what Jeff mentioned. I'm wondering if this committee can provide recommendations to the programs on what types of information should be included in those documents that send -- are sent out from the states to the providers with the lab -- with the newborn screening results, so that -- You know, they can design them however they want, they can use whatever language they want, but that some core elements, which this committee could come up with, should be included in those documents, because those are the documents that the physicians are A) most likely to see and B) most likely to use because they are attached to the results themselves.

DR. JOSEPH BOCCHINI: Thank you.

DR. MEI BAKER: This is Mei. I have a question to ask. What's this different than ACT Sheet?

DR. BETH TARINI: So, the ACT Sheet -- I guess, A) if the ACT sheet is -- I don't know the

answer, because I don't know what the content we want to be in is, if the content differs from the ACT sheet. So, that would be the first thing to look at: Are the ACT sheets currently sufficient in what the Committee thinks should be included on this disclosure -- or on these forms, for instance, A), and then B) if that is -- if they are, then do we recommend that all states use the ACT sheets?

DR. JOSEPH BOCCHINI: So, one of the --

DR. BETH TARINI: Because acting -- because acting -- let me -- let me just finish. I -- I just thought of something. Because they need -- the -- the key is, the information needs to be all together when that, I assume, fax comes through. You cannot expect -- well, you can expect, but I believe you will be sorely disappointed if you think that physicians will go online, in large number, to get the ACT sheets. I think it's -- it's -- it's a matter of, sort of, user design, everything at the fingertips. So, I think if we think the ACT sheets are

sufficient, then we must encourage the programs to include them when they fax out the results.

DR. MEI BAKER: Very good point.

DR. JOSEPH BOCCHINI: The -- the other thing, Beth, just to follow up, is that, certainly, the product that the Education and Training Workgroup has been -- has developed to go along with the ACT sheets, in terms of how to provide information to the parents, certainly is a step in the direction of providing that information that you are talking about. That -- that might help with the just-in-time approach to give what is needed, and I -- I don't know how much that product addresses the kind of things we're talking about today.

DR. BETH TARINI: Yes, and I guess that's the question, that -- that what we've worked on and what the communication of it -- well, what it -- what the content of it is now, does that contain, with the ACT sheets, everything the Committee thinks the physicians should know? Again, this gets to my original point, which is,

we may already have the content and the tools, and they may just need to be tweaked in the content and/or their delivery. So, I -- I think -- I think that the -- but the first step is to assess, what are the core elements of education that have to be given to the physicians at the time of the results.

DR. JOSEPH BOCCHINI: Yep. Sue Berry?

DR. SUSAN A. BERRY: This is Sue Berry. Two comments: I hear about the just in time for positive results. I want to emphasize that one of our elements of education -- and I think it's a very important one -- is, while -- So, what I'd say is, while getting a positive result is a rare event in a physician's or a care provider's life, getting negative ones is not.

And one of the things we wanted to emphasize, as an opportunity to educate the public, is that point of contact as a plan for mediated and understood education about newborn screening: You know, you've got -- you -- your lucky baby has a negative test. We're very

thrilled that your baby is safe and healthy because of this wonderful public health thing. You know, screening isn't perfect. You know, we still need to keep -- be vigilant for your baby's health.

Those kinds of things happen every day in physicians' office, or should, and -- and that's another opportunity to, sort of, raise awareness. So, a -- a -- a -- a critical element in this educational process has to be education about the process itself. I think that's one of the things Mei was emphasizing.

The second thing I'd comment on is that there's a whole new cohort of specialty physicians, who have never really thought very much about newborn screenings, who are suddenly encountering it: the immunologists, who now have to do with -- deal with newborn screening results for SCID, the neurologists who are suddenly now going to have to hear about SMA results. And so, there's another facet of education that I just want to put on the table as long as we're putting

those on the table, although I don't think that's the direct target of the conversation today. I just don't want us to forget that there's a whole new cohort, in addition, that requires some guidance and information about the utility of newborn screening.

DR. JOSEPH BOCCHINI: Thank you. Now we have comments or questions from organizational representatives. I have Bob Ostrander first, then Natasha Bonhomme, and then Susan Tanksley.

So, Bob?

DR. ROBERT OSTRANDER: First of all, when we're talking about an educational initiative to remind physicians who receive test results that there are such things as false negatives, and they should not ignore symptoms, I think it's very important, if we're going to do that, that in the same presentation, we remind people that these are screening tests that need confirmation. If you just talk -- talk about the possibility of false negatives, the logical thought of the person receiving that message is going to be that

all positives are true positives, and that is an equal problem.

And it's interesting: You know, I -- I have been working on educational projects with family physicians, lecturing and -- and sessions, and for some reason, the -- the fact that newborn screening is a screening test -- It seems to be separate in everybody's mind from other screening tests we deal all the time -- with all the time, like mammograms and colon cancer screening, where these principles are fairly obvious. So, you know, again, I think you need to -- if you're going to address the false negatives, you've got to address the -- the fact that these are screening tests and that positives need confirmation.

A second issue is, I just would affirm that we should try to pull all these different efforts together on this and not have a whole bunch of siloed projects that are slightly different, one from the other. The AAFP doesn't have, kind of, a specific thing on newborn

screening that we're disseminating widely as part of certification or anything. I mean, I gave a lecture recently to a national meeting about it, but I -- I think we should work together. And, again, there's a -- we're working on this -- the Education and Training Workgroup is working on this, how to deliver news, and I think a lot of that gets incorporated with this.

The other issue I'm just going to mention in terms of the AAFP is, this is always hard to get on -- on radar, because the family pregnancy community has broad education on genetics and genomics, and when we were talking, I just looked up what the syllabus is for our self-assessment module on genetics is, and it's -- for family docs, an awful lot of it's on the adult stuff: the 23andMe, cancer genetics, family cancer syndromes, and all that. So, you know, on our end of the world, it's going to be a little bit harder push, I think, than for the pediatricians, but I'm willing to continue to lead the charge.

And most importantly -- and I think this

is probably the thing where the Committee should, I mean, exert whatever pressure it can, is -- I agree with whoever it was that said, having this little educational blurb, in a just-in-time format, when the result comes is going to be critical, because we're not going to get traction with publishing white papers or sending out modules and -- or even, necessarily, having people try to go to the ACT sheets when they get the positive results. So, you know, if -- if we could do or facilitate the production of a suggested little blurb that we could disseminate to the state labs and -- and encourage them to include that when they send test results, both positive and negative, I think that's probably where we would have our biggest impact.

DR. JOSEPH BOCCHINI: Thank you for the comments, Bob.

Natasha Bonhomme?

MS. NATASHA F. BONHOMME: Hi, can you hear me?

DR. JOSEPH BOCCHINI: Yes, we can. Go

ahead.

MS. NATASHA F. BONHOMME: Thank you. So, there's been a lot covered that I could speak to, but two pieces, one in terms of the document that could explain, you know, returning results and what they mean. This is something that, through Baby's First Test, our state workgroup has created, and it is currently going through the final stages of review with our Community and Consumer Workgroup, so that it has been viewed by experts on both the technical lab side as well as experts on the family experience side. So, once that's ready, we are happy to share that with anyone and everyone that's interested in it.

And I think that's just an example of the fact that there are so many initiatives and documents and things being created, particularly around education, that having that effort already been done, it'll be really great to see, how do we pull all of that together and also look at it from more of a health communications-communications science perspective.

And what I mean by that is that it isn't just about having the information be accurate but that we also understand how we're going to get it into the -- into someone's hands so that they read it, they digest it, and they know what to do with it. And I think that's a really critical piece and, oftentimes, the piece that people don't spend as much time on but is, kind of, the main thing. It's great to create something, but if it doesn't go anywhere, and the same people just keep reading the same things, then we really aren't moving the needle.

And I think, as we're continuing this discussion, for me, it was really hard to track if we're talking about a media issue, which is one thing, an education issue, which is another thing, or -- or/and are we looking for a certain type of behavior change. Those are all different and really need different approaches. So, depending where the Committee goes in terms of how this is prioritized and how it's addressed, I really encourage you to look at it from that

viewpoint, as well, because through Baby's First Steps, that's really what we've learned in our -- you know, the site's, I guess, now seven years old -- in working on this, that it's about having both the concrete content but also how it's going to get out there and the health communication science behind it, so. And as always, I'm always happy to help with any of those initiatives and bring the experiences that we have into that. So, thank you.

DR. JOSEPH BOCCHINI: Thanks, Natasha. I think I'd like to -- I think that all three of those elements are -- are components of what we need to consider. So, I think that's a really good way to put it.

So, up next is Susan Tanksley, then Debbie Freedenberg, and then Beth Tarini.

DR. SUSAN M. TANKSLEY: Hi, this is Susan Tanksley. Can you hear me?

DR. JOSEPH BOCCHINI: Yes, we can. Go ahead, Susan.

DR. SUSAN M. TANKSLEY: All right. So,

why I originally raised my hand was because I wanted to emphasize that -- like Dr. Berry did, that I think the issue here is more about those results that come back and appear to be normal, or negative, however you want to term them, and the response of the person receiving those results, so the pediatrician, and how that information is understood and relayed to the family then.

The point that Natasha just brought up, and -- and I think Beth also brought it up, was the question of, you know, what is it that we're looking at? Is it the media issue? Is it an education issue, and is the behavior change needed? And I think the answer is, it's all three of those.

So, the reason this issue initially came up was because it was a media issue, and some of the stories that were told, you could -- you could tell from reading them that it was the response of the physicians that could have been different and would have changed the outcome.

And so, therefore, I think that there, you know, is the education issue that we've been talking about, and that is back to, what is a -- what does a normal screening result mean, and what does it not mean? And that behavior change needed is, I think, based on education that would be provided.

DR. JOSEPH BOCCHINI: Thank you, Susan.

Debbi?

DR. DEBRA FREEDENBERG: Morning. Sorry, I have some technical challenges here.

But -- so, most of what I wanted to comment on has -- has already been said, but what I wanted to emphasize is that we hear, over and over again, we invest a lot in education across the whole spectrum of newborn screening stakeholders, and we still have the same concerns and issues as the rest of the country does in the response to the screening test. And we have heard -- even on positive screens, we have heard, over and over again, from various programs and various parents that the pediatrician or the

primary care's response was that: If something's wrong, the lab would have contacted me, or the program would have contacted me, or somebody would have contacted me; so, I'm not taking action on this. You know, for us, we have internal follow-up, and we can handle that, but I've seen it, over and over again, across the country.

So, part of that discussion is truly what a negative test means but -- as well as raising awareness that just because you didn't get a special contact, however your state's running it, that you still have to be cognizant of that child's symptoms or lack of symptoms and that child's risk.

DR. JOSEPH BOCCHINI: Thank you. Now I have Beth Tarini, Scott Grosse, and Mei Baker.

Beth?

DR. BETH TARINI: So, one reminder, and I -- I bring this up often during my E&T Workgroup meetings, is that we are a committee with no funding -- I mean limited, depends how you want

to define the funding, but -- but these projects -- I agree that these are all problems, but sometimes we get a little -- I -- I worry we get a little -- you know, we're going to, you know, resolve world peace. That doesn't mean I don't think we should reach for the sky. I think we should dream large but with realistic expectations that -- that -- that our workgroups and our committee do not have funding to do robust projects.

That being said, I think we could leverage the existing -- and I always say this in the education community -- the same resources, connections, that are rich in this committee and its workgroups, so -- and be very mindful of, again, what we're already doing and -- and how that can be leveraged. And so, maybe, on the heels of this, it would be helpful, at the next meeting, to have the stakeholders from -- who are already engaged in educational activities at APHL, at Genetic Alliance, at CDC, present what is actually the landscape and what we're actually

doing already.

DR. JOSEPH BOCCHINI: All right, thank you.

Scott Grosse?

DR. SCOTT GROSSE: Hi, Scott Grosse. I had suggested to Kellie and the workgroup at APHL that the endocrine disorders could use more attention. Most of the report focuses on metabolic disorders, and I think that's very well done, but the most common disorder in newborn screening is congenital hypothyroidism. And there's a lot of variability across states and how it is done, including one screen versus two screens, because there's a recent MMWR article from Utah which points out that many children are identified on a second screen in that state that would not be identified on the first screen.

And so, more discussion about the sources of variability for the congenital hypothyroidism, I think, would be of great interest, maybe a topic for future discussion. Thank you.

DR. JOSEPH BOCCHINI: Thank you.

Mei Baker?

DR. MEI BAKER: Yeah, I just wanted to quickly share my reflection with this very -- I feel very rich discussion, and that if it can view -- I mean, myself, take-home message for the discussion is, I feel there's not a single way we can accomplish what we want to accomplish. I do feel, due to foundation, it's important.

It's why I feel Sue's work is very important, because that is, in a fashion, non-biased and a very comprehensive medically to help primary care physicians build a foundation, what a newborn screening, how, what -- and false positive/false negative all be comprehensive, but with this, adding on the in-time information -- because we don't expect, at that moment you have screening positive report in your hands, you just would use one sheet. You can have 40 on the standard newborn screening system. So, I think it is combination.

But also, I really think Natasha make so important a point is communication signs. So, we

-- you think about ourself, NewSTEPs, APHL, because we don't have direct connection with the family, direct connection with the primary care physician. We can do the best is create material, but the material you create is not get to the people we want to reach, and I think can be inefficient. So, I -- I just feel like all the pieces are there, how we link them together, and I think will be better to get where we need to be.

DR. JOSEPH BOCCHINI: Thank you.

And then, Natasha?

MS. NATASHA F. BONHOMME: So, hi. I think the one -- two pieces I wanted to bring up is, you know, we did talk about the investment in education, and I would challenge that we invest a lot considering when you look at other large-scale campaigns, it's always the education piece that is the multi-million-dollar, multi-multi-year approach.

And so, we have to think of it from that viewpoint, as well as the fact that education

can't just happen once. People are having babies every day. People are going into the health profession every day. And so, it's something that's ongoing, and so, just putting that within that context --

And then, also, just to reiterate in terms of the point of the provider education: It is really important that we make sure that the messages that are going out to health providers are not just compatible but complementary to the messages that we're putting out to families, because so often, we know that healthcare providers are looking for not just the general concepts of what to say to families but the exact language around it. It's why there is such a high usage of Baby's First Test by health care providers, so we really target towards families.

And so, when we are down the line in thinking about how to get this information out there and how are we going to engage health professionals in making sure they have what they need, to really look at their behaviors of how

they get information, and, again, not speaking about the specialists who are deep in newborn screening but all the people, as I believe it was Sue Berry who was saying, that people who haven't been trained around this, who didn't realize this would be on their plates, but they are because of the conditions that we are now screening for.

So, just a bit more food for thought as the Committee determines what the next steps are and -- and how we will be seeing -- what are the key questions in this we -- you need to address, and how will we be addressing them. Thank you.

DR. JOSEPH BOCCHINI: Thank you, Natasha. I think this has been a really excellent discussion and, I think, gives us much food for thought on -- on next steps. And -- and I think that it's clear that the Lab Standards Workgroup has contributed significantly to the APHL document, as well as to education back and forth with the full Committee, and we're going to see how that document gets finalized, but I -- I think that it is near -- near coming out on it --

on the APHL website. And it looks like QI projects are now going to be coming forward through APHL that will help enhance the -- the efforts that will be made to utilize the resources that are on that document.

And it looks like, from our discussion today, that the focus is really, now, for us, more on, how do we -- how do we educate the media, the -- the parents, the public, and providers on how to get the correct messages out about newborn screening, how to interpret results, and, most importantly -- I think, maybe, Susan Tanksley mentioned this, about understanding -- having providers and parents both understand that newborn screening is important but that a normal test doesn't rule out a condition. And so, that -- I -- and I think that -- that's -- that's probably the most important message, I think, that all providers should learn when they're taking care of patients who come in with acute symptoms.

So, I think we need to continue to have

the Lab Standards Workgroup look at what's coming from APHL and the Education and -- and Training Workgroup continue to evaluate, as Beth indicated, perhaps, first step, maybe looking at the landscape and then coordinating efforts with -- with Natasha and -- and -- and others to try and -- and -- and not work independently, when we can cross-fertilize what -- what we're doing in -- in a variety of different areas. So, I want to thank everybody for a really good discussion, and I think that helps us move forward.

And then, lastly -- this is not directly related, but from what Scott mentioned about the hypothyroidism, the article that he mentioned is included in your briefing book so that Committee members can certainly look at that, and then we can certainly consider that for future meetings to kind of -- to look at that and see how that fits with, perhaps, going forward.

So, that'll conclude this session, and now, we're going to take a short break. I think we are pretty much on schedule, so we'll come

back at 20 minutes after the hour -- that's 11:20 Eastern Time -- and continue with the next presentation. So, everybody take a short break. We'll see you back in about seven or eight minutes. Thank you.

(Whereupon, the above-entitled matter went off the record and then came back on.)

DR. JOSEPH BOCCHINI: All right, welcome back, everybody. This is Joe Bocchini. I've been reminded that each time anyone speaks that even though I've called your name, that you need to give your first and last name for the people who are recording the session.

So, everybody's back on board. Let's continue with the meeting. Next, we have a presentation from Marci Sontag.

DR. CATHARINE RILEY: And this is Catharine Riley. Just, we are having a little bit of technical difficulty. Dr. Sontag, we're trying to pull up your slides now if you can give us just a moment. Thank you.

DR. MARCI SONTAG: No problem.

(Period of silence)

DR. CATHARINE RILEY: This is Catharine Riley again. Thank you, everyone, for your patience while we pull up the next of the slides.

(Period of silence)

DR. MARCI SONTAG: Catharine, this is Marci. We can see the slides online. I'm not sure if there's just a problem in the room?

DR. JOSEPH BOCCHINI: We are not able to see them at the moment, but --

DR. CATHARINE RILEY: So, Marci --

DR. MARCI SONTAG: Okay.

DR. CATHARINE RILEY: -- you're saying you can see them?

DR. MARCI SONTAG: I can see them.

DR. CATHARINE RILEY: Okay, great.

DR. JOSEPH BOCCHINI: Okay. So, just some -- a couple of Committee members, can you see them, as well?

DR. SCOTT M. SHONE: Yes. This is --

FEMALE SPEAKER: I can see them.

DR. SCOTT M. SHONE: -- Scott Shone.

UNKNOWN SPEAKER: I can see them online, yep.

DR. JOSEPH BOCCHINI: Okay. Well, then, we'll go ahead and get that started, and then we'll do our best to try and fix it in -- in the home office here.

So, Dr. Sontag is the Director of the Center for Public Health Innovation at CI -- CI International. She's also the Director of NewSTEPs 360 and will be sharing with us today several videos that have been created by state newborn screening programs in conjunction with the families that they serve in their states. Embedded in Dr. Sontag's presentation are three videos related to timeliness. She will be giving us the story behind these stories, why and how these videos were created, and how they can help us spread the word about the importance of timeliness in newborn screening.

So, Marci, the floor is yours.

DR. MARCI SONTAG: Thank you so much. I'd like to thank you, Dr. Bocchini, and the

Committee for inviting me to speak today. We -- we have spent a lot of time with this committee, talking about the data and what we know about the data from our timeliness efforts, our efforts to improve timeliness in newborn screening, and now I'm going to spend some time today talking about some of the work that's happened behind the scenes that some of you may not know about, some of which is displayed in videos, as Dr. Bocchini has already introduced, and then others -- and some other tools that have been developed by NewSTEPs 360 and our -- the states that we are partnering with.

This slide demonstrates -- or displays an infographic that really shows the complexity of NewSTEPs 360. In the center, you see a clock that is the very little representation of what we're trying to fix here, that timely newborn screening, and in that gray, inner circle there, you'll see 12 icons that represent the activities of NewSTEPs 360 and what we do within this project.

So, starting at 12:00 and then moving clockwise, you see we have quality improvement coaching, we have our online data repository at NewSTEPs.org, the CQI framework with the PDSA study -- study and act cycle. We have annual, in-person meetings that we have conducted at 3:00, technical assistance, financial assistance that's provided directly to the states to help improve timeliness, the monthly all awardee webinars at 6:00, connecting newborn screening to -- programs to partners in other states -- and look at that network that has been developed and the community that -- that has been developed with NewSTEPs 360 -- data visualizations to monitor progress and change -- and you have seen some of those presented to this committee already -- the facilitation of learning collaboratives to identify common solutions at 9:00. We have tools and resources that are both developed by and provided to state newborn screening programs, and, finally, a community for sustainability at 11:00.

On the outer ring, you see some of the ways by which states have attacked the timeliness problem. They're providing education to hospitals, midwives, birthing facilities to help collect newborn screening samples earlier. They're expanding courier services, such that newborn screening samples can arrive to state labs faster, increasing the lab hours so that state labs can test newborn screening samples sooner, and within that, they're also working efficiently within their lab processes and improving those lab workflows to then get those results to the doctors faster. They also are implementing -- implementing HIT systems that help them to share that data in a more efficient manner, such that those results get out, and individuals can act on behalf of that baby, and, ultimately, babies' lives are saved.

Next slide. We all know that newborn screening -- timely newborn screening is critical, and we've spent much of our time within the discussions of this committee talking about

how important that timely newborn screening is, but I want to spend just a moment looking back to where we have come from.

Next slide. We know that this has been -- discussions have been coming to this committee, very driven by parents and advocates, since -- Oh, I'm sorry, can you go back in the -- We were just talking about that one. Thank you so much. And it was the parents' public comments to the Secretary's Advisory Committee, as highlighted here on the Baby's First Test webpage, that really brought national attention to this issue of how important this timely newborn screening is and that while newborn screening has worked so well for most babies, there have been cases in which that timely -- lack of timeliness or a delay in the system has led to a tragic result. So, we really -- we must go back to the advocates and parents who have helped us to recognize this is such a critical problem.

That received some -- those problems received the national attention in the Milwaukee

Journal Sentinel in 2013, and following that and, really, in conjunction with that, this committee continued to talk, and that -- those Journal articles really regalvanized our efforts, and this committee then developed newborn screening timeliness goals that state newborn screening programs are using. At NewSTEPs, we are using those to help set goals for state newborn screening programs and have focused our efforts so we're working toward one common goal across the country.

Next slide. Within that, HRSA developed a funding opportunity, and this was before NewSTEPs 360. This is what I will call a newborn screening timeliness mini CoIIN, and the CoIIN is a Collaborative Improvement and Innovation Network, or a learning collaborative. In this case, it was specifically developed to improve timeliness through the newborn screening system.

Next slide. This CoIIN was run in 2014 and 2015 and was an 18-month effort in which 8 programs participated. We had five members from

each of those state newborn screening programs on each team, and each team was required to include a hospital representative. This could have been a representative from a hospital association or someone who works closely in a hospital on newborn screening and could speak to the challenges of education and collecting timely newborn screening samples in an appropriate way and shipping them in a timely way to the newborn screening lab.

It's important to note for this that there's no direct state funding. While we paid for these states to travel to APHL for in-person training, there was no direct funding for projects. The states did learn continuous quality improvement techniques and were trained on those and then how to apply those to their own local projects.

One of the key elements of any CoIIN -- and one thing we're especially proud of with this particular CoIIN -- is the connections that were made. There was one -- there was one in-person

meeting, as I mentioned, where they received training. Then, there were monthly coaching calls in which our coach checked in with these states and developed a plan for continuous quality improvement. He had monthly webinars, in which all eight programs gathered together to share their successes, share their challenges and failures, and what we learned from each other.

And one thing I can say about this particular CoIIN is, the connection that those eight states made was really -- was really remarkable. And we did that a lot by focusing on virtual engagement: How do we make sure that people are engaged even when on a webinar or on a phone call? And we have learned that the lessons from this really have brought up capability.

So, next slide. With that, I'm going to introduce our first video, and this was from this mini CoIIN, and we asked these eight states that were participating to share their lessons learned with us in a short video that they recorded on their iPhones and sent in to us -- or their other

smartphones. You can go ahead and play the video.

DR. CATHARINE RILEY: Dr. Sontag, this is Catharine Riley. So, we are loading the video. It should start shortly.

DR. MARCI SONTAG: Thank you.

(Video plays)

UNIDENTIFIED SPEAKER: There's no sound.

FEMALE SPEAKER: So, Catharine, we're not hearing any sound on this.

(Video plays)

FEMALE SPEAKER: Keep at it. Be patient and diligent, and never give up.

FEMALE SPEAKER: Number nine: Have a strategy. There are many right ways to approach timeliness. Spend your time on the smart ones that will work within your paradigm.

FEMALE SPEAKER: Number eight: Focus on your high-volume providers first. They can make a big impact on your outcome quickly.

MALE SPEAKER: Number seven: Keep in mind, this is for the babies. Some needed

changes won't affect the outcome data, but they are the right thing to do for the newborn.

FEMALE SPEAKER: Hi, everyone. Number six: Don't forget maintenance. Maintaining timeliness is just as difficult as when you get timeliness started. Don't forget: Maintenance is important.

MALE SPEAKER: Number five: Talking to and learning from other states is so important.

FEMALE SPEAKER: Number four: Find out what is happening in other places. Don't assume you know what other departments are doing, and investigate the current processes.

FEMALE SPEAKER: Number three: Help others understand the impact of timely newborn screening on the families. Don't assume everyone knows why timeliness is important. Start with a why.

FEMALE SPEAKER: Number two: Remember to include all the newborn screening partners within the state that impact timeliness.

FEMALE SPEAKER: To ensure timely newborn

screening, it takes a team and champions from each unit in the hospital, including nurses, lab staff, quality improvement managers, and don't forget the couriers. And speaking of champions--

(Drum roll)

FEMALE SPEAKER: Education and feedback to the partners is key. Once providers are made aware of the reasons for timeliness initiatives, they will run with it. Be prepared for an increase in data requests and technical assistance.

(Video ends)

DR. MARCI SONTAG: So, thank you so much for showing the video. Catharine, we weren't able to hear any sound on the webinar, but this one worked well without sound, and I'd like to give a quick shout out to all of those who participated in this video. And I'm sorry we couldn't hear your voices, but we were able to read what you said. But, hopefully, we can figure out the sound before we get to the last two videos, because I'm not sure they are going

to have the same impact without sound.

So, I will continue on with my presentation --

DR. CATHARINE RILEY: All right, Marci -- Sorry, this is Catharine Riley. Did you say, on your end, you couldn't hear any sound?

DR. MARCI SONTAG: I couldn't.

DR. CATHARINE RILEY: Okay.

DR. MARCI SONTAG: I've gotten some texts, and some of them said they did have sound. I know some people did not have sound. So, I don't know, maybe it's on the --

MALE SPEAKER: No --

DR. MARCI SONTAG: -- computer you have sound, and on the phone you do not. Maybe that's the challenge.

DR. CATHARINE RILEY: Sure. Okay. Thank you for the clarification. So, for the folks -- the Committee members, organizational reps, and speakers -- who are both on the webinar and on the conference line, we did ask you to mute your speakers to -- you know, so there isn't feedback.

So, during the -- the viewing of the videos, for the Committee members, org reps, and speakers, you will have to unmute your -- For anyone on the phone, anyone who's called in, yeah, you'll have to unmute --

FEMALE SPEAKER: Understood.

DR. CATHARINE RILEY: -- to hear.

FEMALE SPEAKER: Mm-hmm.

DR. CATHARINE RILEY: Yeah, thank you, and -- and we will pause --

DR. MARCI SONTAG: Great.

DR. CATHARINE RILEY: -- for that before we view the next video.

DR. MARCI SONTAG: Perfect. Thank you, Catharine. That's --

DR. CATHARINE RILEY: Thank you.

DR. MARCI SONTAG: I'm glad to know that. Okay.

So, we'll move on to the next slide. And so, following up on the success of that mini CoIIN, HRSA announced a newborn screening timeliness quality improvement initiative, and

this was announced at the same time that this committee was finalizing those goals for -- and voting on the goals for our -- our timeliness. So, three years of funding that was announced was CQR10-based (phonetic) and state-based projects, and this time, those state-based projects did come with funding for states. So, they could tackle something that they knew was a challenge within their states and have some funding to support that to make those changes that are needed. So, with 24 programs -- or there currently are 24 programs encompassing 28 states that were funded, and, again, our key to success on this is a collaboration between the states, the partners, and the NewSTEPs and NewSTEPs 360 staff that is working with them.

Next slide, please. As mentioned earlier, each team chose their own focus area, and there's a missing hospital in the upper left corner, but this is the -- focus areas could be: the education within the hospitals, getting them to collect those samples, getting the samples to

the laboratories faster, working on laboratory processes, working within data systems to improve how data is shared, and then, finally, that follow-up process to get those results out to the pediatricians and the subspecialists.

Next slide. And we know that we've seen -- there are -- have been great improvements in timeliness. While we're still not completely there, we still have work to be done, the timeliness efforts that have gone into this -- the efforts that have gone into improving timeliness have really made great improvements in the system, and tens of thousands of babies -- or hundreds of thousands of babies are being screened earlier throughout the country, with those results being reported out in a more timely fashion. You saw those data presented by Mr. Joshua Miller in November of 2017, in a presentation to the Secretary's Advisory Committee, highlighting that interim success. And so, as we know, it takes a village of unsung superheroes.

Next slide. Here's a picture -- or some pictures of those unsung superheroes, and just a few of them, at one of our NewSTEPs 360 in-person meetings. You see them sitting at a roundtable, rolling up their sleeves, and really diving into the challenges of newborn screening and fixing the timeliness problem.

And this is just a small snapshot of the people who are working on this -- this challenge throughout our country. You think of all of the people who are on our phone, the people who are working in hospitals, birthing centers, laboratories, working for the couriers, working in follow-up. There's a network of people working to ensure that individuals -- individual babies that are born with these life-threatening disorders have the best chance through a timely newborn screening.

Next slide. One of the tools that has been developed by this network of individuals is a timeliness toolkit, and we've done this in partnership with the March of Dimes and ASTHO.

And this toolkit has really been developed to help states work towards having expanded courier service and more operating hours, or expanded operating hours, within their state newborn screening programs. And I'd like to give a quick shout out to Sarah McCaffin (phonetic) for her efforts in pulling this timeliness toolkit together. This is available at NewSTEPs.org, and if you haven't already seen it, I would highly encourage you to do that.

Next slide. And now I want to take just a moment to talk about what we do at our -- our in-person meetings. This is our final NewSTEPs 360 in-person meeting. As mentioned earlier and as you saw in the snapshots of people at the meetings, we have opportunities for networking, for sharing ideas and solutions, really bringing state newborn screening programs together. And this is a picture of -- this is half of the room at our last meeting of bringing those people together to share ideas and solutions. Individuals are getting up early, staying up

late, and talking over dinners and happy hours about how we can improve the newborn screening system.

At this particular meeting, we also had a focus on skill building. Some of the examples of that is, we had workshops and breakouts on budgeting, on building the process maps, and on storytelling to share that story of our successes in newborn screening. And what we found is, there are so many successes in our program, and we are working so hard to make those successes happen, that -- that sometimes, we don't have time to share those stories. And sharing the stories of the successes helps to get buy-in from our legislators, the decision-makers within states, from families, from advocates, and it also helps us to celebrate our own successes.

Next slide. So, one of our skill-building sessions was a video-production skill building. Many state newborn screening programs proposed to develop videos specific to their states, so they can share those successes and

stories, or maybe they use those videos for training purposes. And hiring a video-production crew can cost a lot of money and be outside of the budget of a typical newborn screening program.

So, we brought this group in to help demonstrate how to develop an inexpensive video and give them those tools of, what can you do with a video with just your smartphone and a few other tools that might cost $300 or $400 total, and then how do you pull that story, that interview, together with a few simple skills. So, it was about a 45-minute breakout session that helped to arm our state newborn screening programs with those skills.

Within that process, we wanted to demonstrate how this could be done and what those results of a video just done by your iPhone or with a smartphone -- what that could look like. So, this group, Denver Film and -- Denver Film and Digital created a custom video that's thanking newborn screening professionals.

Next slide. Now I'd like to introduce that video, and we played this video to kick off the beginning of our last NewSTEPs 360 meeting as a way to bring everyone together and to say: Thank-you for all the work that you have done. So, with that, I would like to start the video.

DR. CATHARINE RILEY: Okay, Dr. Sontag. So, we're going to pull up the video, and just to remind Committee members, org reps, speakers, and anyone listening in on the phone: If you have muted your speakers for the webinar, if you could go ahead and unmute those, and, hopefully, that will solve the issue of folks being able to hear that. So, we'll load that now.

(Period of silence)

DR. CATHARINE RILEY: Hi, this is Catharine Riley again. (Audio feedback). If you unmute your computer speaker, you should be able to hear the video, and we'll go ahead and get that started.

(Video plays)

FEMALE SPEAKER: Hello?

DR. MARCI SONTAG: Catharine, this is Marci. Can you all see the video?

DR. CATHARINE RILEY: Hi, Dr. Sontag, this is Catharine Riley. Are you -- I think we have some folks that say they cannot hear the video; is that correct?

(Off-the-record conversation)

(Video plays)

FEMALE SPEAKER: My younger son was born in 2012. He was several pounds lighter than his brother and came two weeks early but was still perfectly healthy at birth. When he was three days old, we took him home from the hospital; it was then Friday night. We had a big family dinner, and we'd just finished singing Happy Birthday to the new baby and were cutting slices of cake when I heard my husband's phone ring, and it was our pediatrician who was calling. He had gotten a call from the Colorado newborn screening follow-up team that said our son had screened off the charts for a rare, genetic, metabolic condition called medium-chain acyl-CoA

dehydrogenase deficiency, or MCAD.

FEMALE SPEAKER: It was probably, like, a day or two after she was born. We -- you know, I took her to her pediatrician, you know, and they did her -- her newborn testing. And when I first found out, I was just told that, Your daughter, I'm sorry to let you know, has sickle cell disease; her hemoglobin is not normal by any chance. And that was just the start of our journey.

MALE SPEAKER: We got the phone call, and I wasn't home; his mom was. And then, she called me and said, Something's wrong with Mason. We need to get him in right away; it's urgent. And, you know, of course, I didn't know what to think, and I didn't know that it was from the testing at that point. And so, we took him back in and had to get the retest token (sic) from it, and that's when we found out he has hypothyroidism.

FEMALE SPEAKER: So, Noah died about 10:00 p.m. on June 30th, 2009. And then, we got the call probably about 10:00 the next morning

from the pediatrician. He had heard what had happened and also gotten his newborn screening test results back and realized that the information had come too late.

DR. PETER BAKER II: The newborn screen happens with every -- almost every pregnancy and delivery in the United States, let alone the state of Colorado. It's a test that nobody thinks about, but when it's done the right way, it saves lives. When it's done the wrong way, lives can be lost. And so -- and part of that depends on timeliness.

MR. JOSHUA MILLER: Really, we've seen the biggest, I think, improvement in terms of the time from specimen collection at the hospital to receiving the specimen at the newborn screening laboratory, which is a huge component to improving timeliness.

DR. PETER BAKER II: I try to keep it in mind that each result we get back is a child; it's a life, and it's a life that we can potentially save. And so, I don't take that for

granted, never.

MR. JOSHUA MILLER: It means saving the very thing that probably means the most to every parent out there, and -- and that means the world to me. That -- that makes what we do with NewSTEPs very fulfilling.

FEMALE SPEAKER: I'm -- I'm incredibly grateful for the steps that this community has made towards making this a priority and trying to head off stories like mine from ever happening. That is the way that it should be, and I love their commitment to this, just as -- as strong as mine has been.

FEMALE SPEAKER: We -- I don't know where we would be without these newborn screenings. I think that we would be lost and probably in a lot of hurt and pain.

MALE SPEAKER: Mason, he's normal. He's a six-year-old, he's a handful, and he's -- he's perfect.

FEMALE SPEAKER: Newborn screening made all the difference in our life. We have a child

who grew and developed normally. Thank you for what you do. Thank you for giving families information that they need to keep their children safe. We are grateful for you every single day.

(Video ends)

DR. CATHARINE RILEY: We're going to switch back to your slides now.

DR. MARCI SONTAG: All right, thank you, and I apologize for any technical difficulties. If you would like to watch that video again, I would encourage you to do so. It's on our website, at NewSTEPs.org. And the message from that video -- you know, we had family advocates -- and these are advocates who have been very active. Some of them have come to speak at this committee meeting. Some have been very closely involved in the Baby's First Test Challenge Awards and other advocacy efforts with Baby's First Test. But you, hopefully, were able to hear them speak from the heart and say thank you for all that you do, and thank you for the effort that you put in, and they're really grateful for

all of the work that you do every day.

Next slide, please. So, you can see some of those family advocates here in that last slide, and you might not be able to see the emotions in their faces here. This is at the closing of our meeting at NewSTEPs 360. We invited the families to come and watch a screening of this -- this video, and this was the first time they had seen it. And you could see the emotion in their eyes and the -- the -- the gratitude they have for all of the work that is happening among this community.

So, I want to take a moment and thank all of you on this committee and all of you who are listening on the webinar for all that you do for newborn screening and on behalf of these babies. The -- the army of people that are helping to make the system better is just inspirational.

So, the meeting closed with our final video and -- and a thank you, and these families came in and gave roses to everyone who was in the room to say, Thank you for all that you do.

So, with that, let's try -- this is our final video. Next slide or final video, either one. And as they're setting that up, just remember that there are many families out there, and I know many of you don't actually get to see those families, but they are very grateful for all of the work that you do in newborn screening.

DR. CATHARINE RILEY: Thank you, Dr. Sontag. This is Catharine Riley, again, and we're pulling up the last video, and we will also make sure that these are available on the Committee's website, posted along with your slides, for people viewing, as well.

DR. MARCI SONTAG: Thank you so much.

DR. CATHARINE RILEY: Go ahead and get this started. Thank you.

(Video plays)

FEMALE SPEAKER: To the community that has worked so hard to make this dream a reality: I just want to say, thank you so much from the bottom of my heart. It's incredibly validating that my story was heard and respected along with

other families that have been through similar situations as me. To know that this is something that you value, as well, and are willing to bend heaven and earth to make happen for -- for the children of our country -- So, thank you for -- I know that it's hard, and it's expensive, but you guys have done it and continue to be cognizant of -- of, you know, the importance of timeliness. I just -- Thank you, thank you, thank you.

FEMALE SPEAKER: But we thank you, you know, for everything that you guys have done to try to, you know, make it easier and more accessible for us to know what our children could potentially go through.

MALE SPEAKER: Thank you for doing the newborn screenings. It's changed our lives and given us the son that we have today.

GROUP OF SPEAKERS: Thank you.

MALE SPEAKER: Thank you.

FEMALE SPEAKERS: Thank you.

ADULTS AND CHILDREN: Thank you.

CHILD: And thank you for saving my brother's life.

(Music plays)

(Video ends)

DR. MARCI SONTAG: So, really, very little else needs to be said. Thank you, all, for all that you do.

I'd like to go to my last slide and give a few thank yous, starting with: Thank you to the Committee for allowing us to present this and our work on NewSTEPs 360. We are very grateful to all the newborn screening programs who have joined with us for this effort. I'd like to thank the families who were involved in this video, Denver Film and Digital for making the video, our NewSTEPs CoIIN team, the eight states that initially participated, the 360 state and regional teams, and our teams, both in Colorado and at APHL, who have worked so hard in -- on our NewSTEPs efforts. And, finally, I'd like to thank our funders at HRSA.

Thank you very much, and I'd be happy to

take any questions.

DR. JOSEPH BOCCHINI: Marci, thank you for this presentation. I think the story behind the story is, really, pretty dramatic and really highlights the amount of work that's necessary to make the changes that are necessary to make the system work but also shows us the final outcome, which is what we're all -- what we're all focused on, which is to improve the outcome for the newborn baby. So, thank you. Great.

Let's go ahead and open this for any questions or comments. I think we can spend a few minutes before we go to lunch if anybody has questions or comments for Marci or on what they just saw.

DR. MARCI SONTAG: Dr. Bocchini, while we're waiting for questions, I wanted to also let everyone know that these last two videos have been viewed, kind of, in a similar way to what we have -- we did with our NewSTEPs 360 meeting. And we've had states who have viewed this in their newborn screening advisory committee

meetings to thank them for the work that they're doing, at other stakeholder meetings to really get that word out that it does take a village, and every piece of that village is important. So, it's a -- it's a way for all of you, as well, to be able to use that and say thank you to your stakeholders.

DR. JOSEPH BOCCHINI: Thank you. Any additional comments? Committee members or org reps --

(No audible response)

DR. JOSEPH BOCCHINI: Okay. Well, Marci, again, thank you for --

DR. BETH TARINI: I'm sorry.

DR. JOSEPH BOCCHINI: Oh, wait --

DR. BETH TARINI: Beth --

DR. JOSEPH BOCCHINI: -- we've got --

DR. BETH TARINI: Beth Tarini.

DR. JOSEPH BOCCHINI: Dr. Tarini, remember to announce yourself. Beth?

DR. BETH TARINI: Sorry, I don't want to stand between -- oh, hold on -- between us and

lunch. I just had a quick question for Marci. Marci, is -- is one of your goals that this video could then be disseminated through the programs to, like, hospitals, couriers, things like that, and is that goal -- or -- and if that is so, it could be measured? Because we often get asked, in the E&T group, about measuring education, and, in fact, I was wondering if you might have any tips on that.

DR. MARCI SONTAG: So, we are -- our goal itself was not to do that, but the impact of this video was, really, for the purposes of the meeting and to demonstrate how to develop it, to -- to develop a video using, you know, equipment you have on hand. However, we are now measuring that impact of -- well, we're measuring the number of hits, at least, that we're seeing of this video. It's -- the link that you have is a YouTube link, so we can see how many hits the video has had, but we are not specifically measuring it right now as its impact in hospitals and birthing facilities, but I think that's a

good idea of something that's a way we could be using this at the local level.

DR. BETH TARINI: Sorry, I forgot to introduce myself: Beth Tarini. But a follow-up question, then, Marci, is, so is the video process what you're helping -- is the video, I guess, a consequence of trying to help the programs develop their own videos, and this is just -- and this is the product, and that process is what was discussed at the in-person meeting? And that's one of the main goals?

DR. MARCI SONTAG: So, it was -- it was actually a two-fold process or a two-fold goal, here. So, the first was how to develop a video, both through the technological means and also how to interview people to get those stories out. But we also had a couple of questions on storytelling itself and how important it is to get that message out. And once we can get that message out to the appropriate stakeholders, it helps them with buy-in.

So, the consequence of the video itself -

- We hadn't actually anticipated this video being used as widely as it, perhaps, is being used, which, I think, is fantastic. So, we know how powerful those family stories are, and if you're the person who's delivering the sample on the UPS truck from one place to another, and you know that that sample is related to a baby, and we can share these stories with the -- that UPS courier driver, that can make a difference. And we've heard stories from states where, when they can engage with those -- those people who have the boots on the ground, it can make a difference.

So, I think, what I'm hearing from you, Beth --

DR. BETH TARINI: Yeah, so that means -- I think that, then, your point is made that -- that -- that -- that they are, then, sharing it with these -- that some of the states are sharing it widely.

DR. MARCI SONTAG: They absolutely are.

DR. BETH TARINI: It seems.

DR. MARCI SONTAG: It -- it seems, and now the question is, is there a way that we could be measuring that, and I thank you for playing that out -- because that wasn't one of our initial goals, and yet, maybe there are ways that we could be capturing that beyond just the number of YouTube hits but to be testing what's happening at the state level with this video.

DR. JOSEPH BOCCHINI: All right, thank you. I see no other questions or comments, so once again, Marci, thank you very much for bringing us up to date on the accomplishments of your group, and I think that'll conclude the morning session. We're going to reconvene at exactly the top of the hour, so that's 1:00 my time, so that gives us approximately 45 minutes for lunch. So, enjoy your lunch, and we'll see you back at the top of the hour. Thank you very much.

(Whereupon, the above-entitled matter went off the record and then came back on.)

DR. CATHARINE RILEY: Operator, are we

all set?

OPERATOR: You are, yes. Please, go ahead.

DR. CATHARINE RILEY: Okay, great, thank you. All right, then, I'm going to turn it over to Dr. Bocchini.

DR. JOSEPH BOCCHINI: All right, good afternoon, everyone. Welcome back for the second session of today's meeting. We're going to start with a -- a roll call.

So, Kamila Mistry?

(No audible response)

DR. JOSEPH BOCCHINI: Mei Baker?

DR. MEI BAKER: Here.

DR. JOSEPH BOCCHINI: Susan Berry?

DR. SUSAN A. BERRY: Here.

DR. JOSEPH BOCCHINI: So present.

Jeff Brosco?

DR. JEFFREY P. BROSCO: I'm here.

DR. JOSEPH BOCCHINI: And I think we have Carla Cuthbert this afternoon.

DR. CARLA CUTHBERT: I'm here.

DR. JOSEPH BOCCHINI: Kellie Kelm?
DR. KELLIE B. KELM: Here.
DR. JOSEPH BOCCHINI: Joan Scott?
MS. JOAN SCOTT: Here.
DR. JOSEPH BOCCHINI: Cindy Powell?
DR. CYNTHIA M. POWELL: Here.
DR. JOSEPH BOCCHINI: Melissa Parisi?
DR. MELISSA PARISI: Here.
DR. JOSEPH BOCCHINI: Annamarie Saarinen?
MS. ANNAMARIE SAARINEN: Here.
DR. JOSEPH BOCCHINI: Scott Shone?
DR. SCOTT M. SHONE: Here.
DR. JOSEPH BOCCHINI: Beth Tarini?
DR. BETH TARINI: Here.
DR. JOSEPH BOCCHINI: And Catharine Riley?
DR. CATHARINE RILEY: Here.
DR. JOSEPH BOCCHINI: For the org reps, Bob Ostrander?
DR. ROBERT OSTRANDER: Here.
DR. JOSEPH BOCCHINI: Debbie Freedenberg?
DR. DEBRA FREEDENBERG: Here.

DR. JOSEPH BOCCHINI: Michael Watson?
DR. MICHAEL S. WATSON: I'm here.
DR. JOSEPH BOCCHINI: Britton Rink?
(No audible response)
DR. JOSEPH BOCCHINI: Jed Miller?
DR. JED MILLER: Here.
DR. JOSEPH BOCCHINI: Susan Tanksley?
DR. SUSAN M. TANKSLEY: I'm here.
DR. JOSEPH BOCCHINI: Chris Kus?
DR. CHRIS KUS: Here.
DR. JOSEPH BOCCHINI: Natasha Bonhomme?
(No audible response)
DR. JOSEPH BOCCHINI: Siobhan Dolan?
DR. SIOBHAN DOLAN: Here.
DR. JOSEPH BOCCHINI: Cate Walsh Vockley?
MS. CATE WALSH VOCKLEY: Here.
DR. JOSEPH BOCCHINI: Shawn McCandless?
DR. SHAWN MCCANDLESS: Here.
DR. JOSEPH BOCCHINI: Great, thank you, all, very much.
So, for this afternoon session, first, public comments. As you heard earlier, we have

not received any requests for public comments for this meeting. However, I just wanted to acknowledge, at this time, that the Committee did receive a petition on June 24th, signed by over 2,000 individuals, supporting the addition of SMA to the RUSP.

So, now we'll turn to our workgroup updates. And so, we will begin with the Education and Training Workgroup, and we have Beth Tarini, Chair of this Education & Training Workgroup, who will present this, and for those of you who are not aware, Cindy Powell has agreed to serve as Co-Chair of this committee, as well.

So, Beth?

DR. BETH TARINI: Thank you, Dr. Bocchini. So, my name is Beth Tarini for the -- the notetaker, and can you check the next slide?

I want to acknowledge all of our members, and you see them listed here. Thank you to all of them for their contributions and participation.

Next slide. And to echo Dr. Bocchini, I

want to welcome Dr. Powell, who is our new Co-Chair, and thank -- and give a thank you to Cathy Wicklund, our former Co-Chair, for the work she has done for us.

Next slide. So, I'm going to go over the two current projects and then the brainstorming we did in our meeting, which was last week. So, the communication guide -- this is the guide that was mentioned earlier. We're in the final stages. We discussed getting feedback from pediatric residents and genetic residents -- genetics residents on the guide. We are working to link it to the ACT sheet, and we discussed identifying the most effective ways to get it to the states, especially the end user. And so, we talked about listservs utilized by members of state programs, APHL, options through the media, and tried to decide and identify other ways.

Next slide. The education guide -- this is the guide that, to briefly review, points out elements of education for newborn screening, or content domains, if you will, and how they might

map to areas of interest for various stakeholders. So, it is also in its final stages and will be posted on the Committee's website. We are trying to identify a way to validate and evaluate the guide, as well as have a way to track or monitor usability, talking about, do we talk about the number of visits to the page, downloads, and also find a way to collect the feedback, either via SurveyMonkey or at the time of the use or, perhaps, asking them to download and then following up a week later. So, we will -- we are working to identify which is the most effective way to solicit the feedback from users.

We also talked about --a -- a good point was made about finetuning the introduction to target the end user, which would include -- which would involve including more background and direction about the -- the utility of the guide. And so, we're in the -- in the process of drafting that revised introduction.

Next slide. And so, we talked about two issues that came up in the Co-Chair's meeting

that we touched on earlier, and those are the issues of -- well, this is -- the project label is -- is nebulous. Provider education was under the rubric of provider education and talked about issues with communicating the limitations of newborn screening was the problem we were seeking to address, and the "who" we talked about were the providers, generalists and specialists, particularly educating them so that they could better inform the families. And we talked about one target, how it would be, perhaps, the North American Metabolic Academy for the specialists.

Now, let me just pause to say, this is not a proposal of a project. The goal -- and I should have started with this in discussing this with you -- is to come to the Committee to solicit thoughts and feedback on what might be, from the Committee's perspective, most useful. We did some bit of brainstorming and then are bringing that small bit to the Committee. And we realize that we have only a small window of time to discuss it today.

Next slide. And public education -- this was also touched on earlier -- explaining that a negative screening does not mean you do not have the disease and that that problem targeting "who" -- public, prenatal, postnatal, child, adult, depending on what is our actual intended behavior change or outcome -- and the challenges we discussed were that newborn screening is complex and nuanced. For instance, we don't want, in citing limitations, to detract from the value of newborn screening. However, we also discussed how this wheel has already been invented. This is done routinely with mammograms. And, of course, there are some hazards, but we could learn from other communities on this. And the target, "how," was, could we get focus groups of parents involved in this issue is one potential idea.

But I will say that after -- of course, these slides were made before today -- after discussion today, I will put in my plug, which does not represent, necessarily, that of the

Committee, which is, I think the first step is -- in any of this is to, perhaps, identify what we are currently doing and what is currently out there and start with a landscape assessment and then -- and then, potentially, move forward with ideas. Because, for instance, Dr. Berry's MOC is, I think, an incredibly valuable addition to the educational landscape.

And with that, next question -- next slide, I mean. I will open it up for questions and/or comments.

DR. JOSEPH BOCCHINI: Thank you, Beth. It's a good summary of -- of activities going on within the -- in the Education and Training Workgroup and, certainly, shows the beginning of the discussion of -- towards the education, perhaps, of providers, as well as the public, related to the issues in -- in lab testing results and screening.

So, let's go ahead, then. We'll start with Committee members who have questions or comments. It's Sue Berry and Cindy Powell.

So, Sue first.

DR. SUSAN A. BERRY: This is Sue Berry. I just want to clarify: It's not my -- my personal MOC4; it's part of the Midwest Genetics Network's, but I appreciate the -- the endorsement, and we will certainly keep the Committee apprised of that.

I really wanted to mention one potential forum for sharing a document like the one you've described, and that's to reach out to the AAP News, which is -- it bills itself as the official news magazine of the American Academy of Pediatrics, and it produces short articles of general interest to pediatricians. While I know that -- that -- that the target's pediatricians, it still could be a -- a resource. And so, it's, perhaps, worth considering reaching out to them for a document as valuable as -- as the communication guide.

DR. JOSEPH BOCCHINI: That's a great suggestion, and I think it is one way to get to a large number of -- of primary care pediatricians

and subspecialists.

Cindy Powell next.

DR. CYNTHIA M. POWELL: This is Cindy Powell. Thank you. And I certainly appreciated the discussion this morning about the -- you know, educating providers, as well as the public, and echo the summary from Beth that, you know, we -- we really need to look at what's already out there before we can really, you know, think about moving forward with any of this.

I think one thing to keep in mind, too, in the -- the groups that were discussed this morning is also that, you know, the -- those, in addition to family practitioners, who care for adult patients -- It's also going to be important to educate the internal medicine folks, because, you know, with some of these new conditions, most cases that are being identified in children, you know, will not have onset until adulthood, if, even, then, but, you know. So, I think that's an important group that we need -- that we should not forget about.

And then, also, for -- I think it's great, you know, what Sue Berry brought up about the MOC and also thinking about, you know, the -- the questions that are on the medical student national board exams and whether, you know, that they cover or include things about newborn screening could be one way to make some inroads. They often reflect some of the goals and objectives for various residency programs. And so, you know, you could work with the -- the boards, as well as representatives from the residency training programs, about how to make sure this, you know, gets into the curriculum, which is already so crowded with other things that students and residents need to know about. Thanks.

DR. JOSEPH BOCCHINI: Thank you. Next, Mei Baker.

DR. MEI BAKER: Okay, yeah. This is Mei Baker, and a couple comments. I just want to, kind of, more emphasize what I feel important is the education. Beside education, also with a

mind to provide tools to primary care physician. Also, I do believe all the education material have some kind of connection link to continued education credit will be -- will be part of a motivation get primary care physician willing to do that.

Another thing that we talk about, ACT sheet and this in-time information -- some point, I think we may think about the combination and so you don't give so many different set material to primary care physician at the time but somehow can do some combination material. So, maybe is another avenue to do so.

DR. JOSEPH BOCCHINI: Thank you.

Next, Shawn McCandless.

DR. SHAWN MCCANDLESS: Hi, this is Shawn McCandless representing the Society for Inherited Metabolic Disorders. Beth, I appreciate the -- the mention of the North American Metabolic Academy, which is a -- a function of the SIMD. That -- the -- the limitations there are that it reaches a very small number of -- they are key

people, because they're trainees in the field of medical genetics, but it reaches a very small number of people once a year.

Also, the curriculum is highly structured and professionally developed and not so easy to change and add to. So, I -- I'm trying to pull up the newborn screening curriculum now and having some problems with the website, but I -- I -- I -- I feel like that's maybe not going to be a very effective mechanism for getting the message out about the -- particularly about the importance of -- of screening as a screening test and not a diagnostic test.

The -- the -- I do think, also, that that population, the -- particularly the people with metabolic training in particular but also geneticists in general, are probably fairly savvy to the idea already that screening tests are not definitive and that they -- when they're asked to see a patient, a normal newborn screening test doesn't rule out the possibility of one of these inborn errors of metabolism. That's all I have

to say.

DR. JOSEPH BOCCHINI: Next, I have Sue Berry.

DR. SUSAN A. BERRY: Yeah, so just wanted to make sure that everyone knows this is not -- we'll be happy to provide information about the -- the details in the MOC4 so that we can make sure that all of the efforts that we do on a regional basis through our HRSA group are also coordinated carefully.

We had planned to incorporate discussion of the communication guide in the MOC4 about -- the -- the module about giving back positive results, because that's one of the intents for that, but we're hoping to be able to, kind of, build on the work that the Committee's already done on the document.

And the other thing I'm just going to throw out there to give a little bit of a shout out to my own Department of Health, which is that Amy Gaviglio and a medical student that I was working with assisted in developing a -- a guide

for our local pediatric practices to give when they are conveying negative newborn screening results. It's -- you know, it's -- we did some vetting with families, and we did our own internal vetting, but it -- it could certainly use some additional work. But it might be a place to start from that I think people would -- our -- our goal was to have a simple sheet that is equivalent to the one that's given out after hearing screenings, when you -- when you don't get referred or when you have a heart screening and your heart test is normal. It's the same thing. And so, it's kind of a nice little one-page document, and if people are interested in that, we can certainly make that available.

DR. JOSEPH BOCCHINI: Thank you. I -- I think that we've had some discussion and presentation of that in the Education and Training Workgroup. So, I think that would be good to continue to share about.

DR. SUSAN A. BERRY: Great, thank you.

FEMALE SPEAKER: Debbie Freedenberg.

DR. JOSEPH BOCCHINI: Oh, next, we have Debbie Freedenberg.

DR. DEBRA FREEDENBERG: So, I really -- This is Debbie Freedenberg. So, I really appreciate the discussion this morning related to education, as well as Beth's comments from the Committee -- the Workgroup, excuse me.

So, I think one of the things that we need to consider as a group is that the way -- if we're aiming for professionals and for pediatricians and family practice, the methodology of how they're learning in education is changing, and like a vast majority of folks will go to an instantaneous, just-in-time pop-up app, or the EMRs automatically link them to a pop-up with information about whatever it is. And so, I think we need to start thinking, also, into the future because we -- you know, as a state, we do the brochures, we do the webinars, we do the talks, we do all of that -- you know, active, facted information, and we pair with our pediatric society and try and pair with our

OB/GYN, which we're not so successful with -- society, which we're not so successful with, but I think we need to start thinking about the way information is now being processed and learned.

And the families, most of them, are younger families, so we're having kids who are also in that same learning paradigm, where, you know, they're much less likely to pick up some printed material than being able to click a button on their iPhone or their Samsung whatever, on their cellular phone or their iPad or wherever they are, you know, their laptops.

So, I think we need to recognize that the educational paradigm has been shifting a bit more towards electronic. And, you know, we've often spoken about, could we just develop an app and put it on there and have everybody pull up what they need. But there are significant barriers to that for us, as well, but I kind of would like -- just wanted to point out that there are newer methodologies that people are using to get information.

DR. JOSEPH BOCCHINI: Thank you. Any additional questions, comments?

(No audible response)

DR. JOSEPH BOCCHINI: If not, I think it's clear that the Education and Training Workgroup's going in the right direction and that we look forward to additional discussions. And did we give you enough to have additional discussions, Beth?

DR. BETH TARINI: Yes, I think that the one piece to tie up is -- is, if this is going to be an interdisciplinary work project, whether or not that is the intention, and then what would be the infrastructure or formal, sort of, sub-workgroup collaboration for that or the idea or intent that we as the Education Workgroup take this suggestion from labs and -- and run with it, if you will.

DR. JOSEPH BOCCHINI: I would -- I would have you take the lead and run with it but then use the expertise of the laboratorians for specific information related to test

interpretation, et cetera, and -- so that working together, we can come up with clear guidance and definitions and -- and interpretation.

DR. BETH TARINI: Okay. Yeah.

DR. JOSEPH BOCCHINI: All right. Thank you very much.

Next on the agenda is the report from the Laboratory Standards and Procedures Workgroup. Kellie Kelm has already discussed, in detail, a significant amount of the work that this workgroup is doing. So, we'll see if Kellie and/or Susan have any additional information to bring forward in their report.

Kellie?

DR. KELLIE B. KELM: Yes. So, as we said, most of what we discussed in our -- in our workgroup meeting I captured in our earlier presentation. And one thing that was touched on briefly, also, in our discussion, that we were -- we -- we also discussed was the recent publication, I believe in MMWR, on one screen versus two screens, looking at the effectiveness

of screening for congenital hypothyroidism, and that's something that we have discussed before and I think might be a -- a future topic that may come around again.

I didn't have anything else. I don't know if Susan's available, if there's anything else that she wanted to add that -- that we discussed.

DR. SUSAN M. TANKSLEY: This is -- this is Susan Tanksley. I don't have anything to add. Thanks, Kellie.

DR. JOSEPH BOCCHINI: All right. I want to thank --

DR. KELLIE B. KELM: All right.

DR. JOSEPH BOCCHINI: -- the -- I want to thank this workgroup for the -- for the efforts that they have made related to risk assessment and -- and then the recommendations that came forward for us to evaluate today.

Are -- does anyone have any additional comments for the Education and -- I'm sorry, the -- the Laboratory Standards and Procedures

Workgroup before we move on?

Dr. Tarini?

DR. BETH TARINI: Hi, Beth Tarini. I have a -- a -- a question or issue to raise, and that is an issue that's come up in some of the newborn screening circles about congenital hypothyroidism and the potentially increase in the diagnosis of congenital hypothyroidism, as well as --

So, this is a little bit -- So there's the screening issues, which are the age-appropriate cut -- age-adjusted cut-offs issue, as well as the diagnostic line for -- for congenital hypothyroidism in the short-term follow-up, and -- and then the overall notice in some states of an increasing rate of congenital hypothyroidism diagnoses. And I'm wondering if this might be an opportunity for cross-collaboration of all three workgroups on this issue and wondering if anyone else has -- has thoughts about that, particularly the lab group.

DR. JOSEPH BOCCHINI: We certainly can

open that up for discussion.

FEMALE SPEAKER: Mei Baker.

DR. JOSEPH BOCCHINI: All right, Mei Baker first.

DR. MEI BAKER: Yes, hi. I just want to -- Oh, this is Mei Baker, and to follow Beth's comments, I feel that maybe need a multidisciplinary, because that -- at newborn screening timepoint, the elevation, and I think, beside of what you said, also the transient, and that's what we need to assess, right? I feel like maybe -- I don't know if we can call this long-term follow-up or not, because I feel this cohort need to be assessed at least for three years, and so we kind of have a sense in terms they're transient or, you know, true cases. So, then, we can better assess the incident rate.

So, almost I -- what I'm suggest that is maybe Laboratory Workgroup and short-term follow-up or even long-term follow-up kind of come to some kind of procedure and recommendations and see each state interesting follow this. Then, we

can collect data at certain time points.

DR. JOSEPH BOCCHINI: So, Beth Tarini?

DR. BETH TARINI: Hi, this is Beth Tarini. Mei, you bring up an excellent point. And, in fact, I think that the point -- the issue of the three-year follow-up to see, does the child actually have the diagnosis we thought they had, or was it transient or was it -- is it -- was it transient or was it permanent, actually, I think, is an example of where Long-Term Follow-Up has -- and I don't want to speak for that group, but Long-Term Follow-Up has a real impact on screening. We often talk about, what's the role of Long-Term Follow-Up. Here's an example where Long-Term Follow-Up actually is -- has a critical role in determining how we diagnose -- how we create and diagnose off of these tests in -- for congenital hypothyroidism.

DR. JOSEPH BOCCHINI: Other comments or questions?

(No audible response)

DR. JOSEPH BOCCHINI: So, I think there's

enough new information that this should be a topic for us to pursue, and perhaps we could put together some presentations for the next meeting to, kind of, charge the -- the three different workgroups, following that -- those presentations, to consider aspects of this that might be things to evaluate or consider to bring back to the full Committee. So, we'll see how quickly we can organize having some presentation and discussion on this topic, so. Thanks.

Okay, are there are any other issues related to Lab Standards and Procedures?

(No audible response)

DR. JOSEPH BOCCHINI: No. Thank you. Okay, the next is the Follow-up and Treatment update -- Workgroup update, and Chris Kus is going to lead this presentation.

So, Chris?

DR. CHRIS KUS: Thanks, yeah. It's Chris Kus from the New York State Department of Health. I'm actually in the Division of Family Health -- it's missed on that slide -- and I'm pinch

hitting for Jeff.

Next slide. Just shows the Workgroup -- current Workgroup members

Next slide. So, this has been a busy workgroup and report on some of our activities. The first one, the medical foods, which completed the report, which was sent to the HHS Secretary as informational, it's going to be posted on the Committee's website and submitted for publication in a peer-reviewed journal is planned.

Second thing, the work we've done in terms of quality measures, the quality measure report, which is completed, and it's, again, going to be posted on the Committee's website. It'll take a little time -- I think in about two weeks, it should be on the website. The plan is to identify journals to publish the executive summary in, not peer-reviewed journals, to consider topic-specific articles that build on the report, consider what aspects of the report tie into the roadmap project, and we'll get into more of that discussion.

I think Bob Ostrander highlighted that there's -- we've had a -- a lot of discussion about a roadmap for long-term follow-up, and there are a variety of audiences that may be interested in the report. So, we're asking the Committee, is there ways that we should do outreach to make sure that it gets to the people that need to see it.

Number three is a long-term follow-up roadmap. We're in the development phase, and we're going to consider next steps based on the Kemper and Lam's Environmental Scan and think about what different components need to be.

Having said that, next slide. A big part of our discussion at our last meeting was about a long-term follow-up roadmap, ideas for moving to an informatics paradigm. At our meeting, Joe Schneider and Bob Ostrander presented some ideas for improving care by creating an integrated system for quality measures, and Joe put out a word that I had to look up, that -- calling it a core desideratum, and -- which essentially means

something that is needed or wanted. And the thoughts we have are, thinking about single registry and QI programs per disease or disease group, considering data consistency as we look at follow-up for different conditions, the idea of a hub-and-spoke data collection system -- my phone answered one of my questions, I guess -- a -- a patient identification and continuity, and coordinated research/QI funding.

Next slide. So, I'm going to throw out some questions for the group, and I'll just read through the questions, and then we can have a discussion. How can data registries inform quality measures that can help us improve informed treatment and follow-up? Is there an interest in creating an integrated system for quality measures? Do we need a core set of data elements that are both disease specific and applicable across diseases? And how can we ensure newborn screening conditions have a follow-up plan?

We had some discussion about the idea, as

people submit new conditions to be on the RUSP, should they have to submit some type of follow-up plan relative to the condition. And the other thing I'll add is, I started to think about this, because we are talking about developing some type of system that reports on newborn screening nationally, and the question is, who's the responsible party for doing that kind of report, and what kind of report might that look like.

So, I'll stop here and say, Bob and Joe, answer some of the questions as they come up, and I'll try to, too. Thanks.

DR. JOSEPH BOCCHINI: Thank you, Chris. Bob Ostrander already indicated he would like to make some comments.

Bob?

DR. ROBERT OSTRANDER: Hi, thanks. I don't know if Joe is here, because Joe really is the lead on this. I just threw some ideas in. But I want to flesh out some of our thoughts about the roadmap, because I think we need to get the Committee guiding us in -- in thinking about

some of the concepts.

I'm going to start with the most virtual one rather than the most important one, and that is our notion that a follow-up and treatment plan is an integral part of a condition be on the Recommended Screening Panel, because if we put conditions on the RUSP, without a system either in place or the architecture for -- the architectural drawing for it laid out, we are really not meeting all of our obligations as a committee, 1), and 2) it puts us behind the 8 ball for reporting back to the Secretary when an approval letter comes back, as it did this time, and I think quite appropriately, wanting follow-up information in 2 years.

And if we don't have a blueprint on paper when we approve it, we're -- we're not very likely to even be able to build a follow-up -- assessment plan in the two years, much less gather any data. So, I think -- we think that it's important, some of us, but we also worry that it's going to be perceived as a barrier to

conditions being placed on the RUSP.

And the other thing that we talked about, and this comes up over and over and more and more at our committee meetings, is that we need to think about what to do about conditions that are already on the RUSP. And we've talked about looking at ones that might need to come off the RUSP, but I think another part of what we're (audio cuts out) is, you know, assigning -- you know, setting a time target to have one of the standardized approaches to follow-up data collection in place for conditions that are already -- already on the RUSP within a matter of, you know, two, three, five years. We can kick some numbers around. So, that's one piece.

And then, the other piece I just want to flesh out the vision a little more for folks to comment on, and we talked about this a little at the last meeting when I brought it up, and that is, you know, working toward a notion where the centers have a responsibility, as owners of registries, for their various diseases.

And then the third component is the notion that we have some standardized measures that will cut across all heritable diseases or even (audio cuts out) things sort of like the medical home issues, and then, you know, an up-front notion about information for each disease or group of diseases that would be standardized across centers. And if we could do this and make this our system, we wouldn't have the polyglot we have now of some of the registries being kept by disease groups, some of them just individual states, and so on.

So, that -- those were our initial vision things. As I said, the -- the Subcommittee's going to flesh some of this out, but I think it would be helpful up front to hear from the Committee if they think some of these ideas are okay and, specifically, the notion of asking candidate conditions to have some kind of architectural drawing in place for how they envision long-term follow-up over the next two to five years.

DR. JOSEPH BOCCHINI: Joe, I think this is a timely question in the sense that our plan is to, kind of, not only review the entire evidence review but also to consider what needs to be in it for us to then make a decision about a condition going onto the RUSP. So, I think it's timely to include this consideration.

So, I have two Committee members: Sue Berry, Jeff Brosco.

Sue first.

DR. SUSAN A. BERRY: Thank you. This is Sue Berry. I love the attention to having us consider the whole system, not just the test, which includes the -- the long-term follow-up.

I will point out work that's been done by the Newborn Screening Transitional Research Network to create the Longitudinal Pediatric Data Resource, which, as a element in its development, created a set of common data elements that were, hopefully, to be used across diseases. While that was developed in a research setting, they were developed by clinicians for assessment of

patients with these conditions because the only way to get some of this information has been in a research environment. And so, a lot of work was also done in working with public health to take some of those common data elements and select appropriate ones that might be suitable for use in public health. And I'm -- I'm hoping some of what Alex will end up talking about will reflect some of -- some of that work.

The one thing I would point out, after running our project to try and do some of this long-term follow-up data collection, is that this requires time, which I would say equals money. Centers will not be able to do the -- even the most cursory sustained follow-up without some kind of support. So, that'll have to be an element that's considered in anticipating a truly comprehensive follow-up system. But I think you all know of my incredible enthusiasm for any plan that includes improvements in our long-term follow-up so that we can realize the promise of newborn screening. So, I'm really excited that

this is a direction we're taking.

DR. JOSEPH BOCCHINI: Thank you.

Now, Jeff?

DR. JEFFREY P. BROSCO: Thanks, this is Jeff Brosco. First, I want to thank Chris for -- for pinch-hitting for me today. I had a -- a conflicting meeting, which, of course, was canceled just in the last two days. So, I'm actually able to be here.

And then, it was great to hear Bob sort of lay out some of the really interesting ideas that he and Joe have -- have proposed, and just want to -- it would be great to hear some feedback now, but I just want everyone to recognize that these are, sort of, the first time our -- our workgroup is speaking about them, and a lot of what we're going to do is -- is actually based on what we're going to hear in a few minutes from Alex and K.K., about, sort of, where long-term follow-up is right now. Their environmental scan's going to be very helpful as we put together the so-called federated system.

We expect, over the next two months at our workgroup meetings, to take what Alex and K.K. have put together, imagine and start drawing out what this federated system would truly look like, and may or may not include some of the elements that -- that Bob just mentioned.

And, just, quickly, Sue, you weren't -- weren't on the last call, but -- but Amy Brower was there, and I asked Amy to talk a little bit about the Longitudinal Pediatric dataset elements and so on. We're -- so, we're well aware of that. The Workgroup is including that, and the question is, how well do those translate into clinical situations, and how flexible are they, and all that sort of stuff, which will be one of the key topics in our upcoming workgroup calls. I think I'll stop there.

DR. JOSEPH BOCCHINI: Thank you. Next, I have Shawn McCandless.

DR. SHAWN MCCANDLESS: Hi, Shawn McCandless representing the SIMD. I -- I just -- I -- I want to reemphasize what Sue said about

the -- the burden and cost to providers, particularly the members of the SIMD, who are -- often, are the ones that are required to enter data into databases. We're -- I -- I am sure our membership will be enthusiastic about a process to standardize and collect meaningful data in a -- - in a standardized way, especially if that reduces the number of different places that we're expected to enter data regarding newborn screening follow-up.

DR. JOSEPH BOCCHINI: Thank you. Next is Chris Kus.

DR. CHRIS KUS: Yes. I -- I guess, as I thought more about this, is, if we move toward this federated system, one of the questions that comes up, for me, is, who will provide the leadership for that federated system, and then comes up the -- the question is, where will the resources come from, thinking -- You know, we have state resources, federal resources, and if you're moving in this added resource needed, where -- where will that come from is a question.

DR. JOSEPH BOCCHINI: I have Scott Shone and Jeff Brosco.

Scott?

DR. SCOTT M. SHONE: Hi, this is Scott Shone. So, I -- I -- I would like to suggest that the -- the -- the -- the planning for long-term follow-up needs to come before a disorder is proposed for the RUSP but be part of these pilot studies that we should be doing and the data that should be gathering in support of the evidence review for a RUSP nomination. I mean, the -- the -- the current system provides no guidance or funding during pilot studies to help collect this data.

And so, while the pilot studies that have been done have been incredibly successful at demonstrating that screening can be performed, these children can be identified, and we can run confirmatory analyses to show that they either have mutations or other biochemical markers indicative of a disease, what is been clinically lacking is the -- the -- the conclusive

demonstration that early identification and treatment leads to long-term benefit. Now, the definition of "long-term," I know, depends on the eyes of the beholder, but I -- I think that we should be -- we should be shifting the inclusion of this long-term follow-up plan and data collection well before it even gets to the Committee but as part of recommendations to researchers that they be included as part of their analyses for new newborn screening disorders before they even make it to us.

DR. JOSEPH BOCCHINI: Jeff Brosco?

DR. JEFFREY P. BROSCO: Yes, Jeff Brosco. Thank you, Scott, that's very helpful, and -- and, really, it helps our workgroup continue to move in that direction, because I think you're right. A lot of this needs to get done at the -- at the pilot stage.

One quick comment, just to make sure everyone -- I'm -- I'm, sort of, channeling Nancy, who would typically be saying this at this point, that a lot of what you've heard may have

been about quality measures and data and quality improvement, and just to make sure everyone understands: This is all in the interest of treatment, because that's really what we -- we want to make sure happens in the long term, and this is just ways of making sure it happens.

And just a last thing for folks who may not have heard these terms before: When -- when we've said "federated system," what we mean is, we don't think it's possible, at least in the U.S., any time soon, to have a single dataset for every single condition that all states and all groups participate in. We just don't think that's realistic. So, a federated system means that, okay, in CF, it may be the CF Foundation that funds and provides the structure for long-term follow-up for cystic fibrosis, but for sickle cell anemia, it might be something else, and for SMA, it might be a different group. And it may be federal. It would be great if it were, you know, national, but we recognize that some things will be patient-registry oriented or maybe

more localized.

So, when we say a "federated system," we mean having as strong a system of long-term follow-up and treatment as possible, probably condition specific, and it's going to -- it's going to be varied and together in a federated way that makes up some kind of quilt that shows long-term follow-up and treatment's being done. So, just for folks who hadn't really thought about that recently, that's the way we're thinking about it now, and we hope to learn what the -- the strand of that quilt will be from Alex and K.K.

DR. JOSEPH BOCCHINI: That sounds good. Additional questions or comments?

(No audible response)

DR. JOSEPH BOCCHINI: All right. I want to thank everybody for the feedback. I think each of the workgroups has gotten feedback that's necessary for them to continue moving forward with their projects and tasks. So, thank you, all.

Next, we have two presentations by Alex Kemper. The first is on -- is a report on long-term follow-up in newborn screening -- so, this is a landscape environmental scan that Jeff was talking about -- and then we'll discuss that and -- and then we'll -- we'll have a second report on the -- the technology -- the -- the evolution of technology in -- in newborn screening.

So, Dr. Kemper is Division Chief of Ambulatory Pediatrics at Nationwide Children's Hospital, and he leads the External Evidence Review Workgroup for the Committee. His group has been working on two reports for us over the past year.

The first is this report on the status of long-term follow-up in newborn screening. It's a report designed to identify knowledge gaps and potential needs. The Committee will hear from Dr. Kemper for the first time today on this topic, although he and his team have been collaborating with the Follow-Up and Treatment Workgroup, as you heard, as their missions

overlap.

And then, the second report is one that he and his team have been working on that outlines the various advances in technology that have already had or will be expected to have an impact on newborn screening. He presented the preliminary findings of this report last year, and today we'll hear an update.

So, let's hear the first report. Turn it over to you, Alex, for the Long-Term Follow-Up --

DR. ALEX KEMPER: Fantastic, and can you hear me okay?

DR. JOSEPH BOCCHINI: We can hear you well.

DR. ALEX KEMPER: Fantastic. So, this has been a great day. I -- I really appreciate the organization of today's webinar because it really builds on all of the things that we've been thinking of. So, I -- I've really enjoyed all the previous presentations, and I -- I hope that this adds nicely to it.

So, I'm going to talk about the work that

we've done in terms of our horizon scan. I'm going to keep this at a relatively high level and hope, to the best that we can, that we can facilitate a conversation on this webinar. It's not necessarily our purview to come up with specific recommendations, but I think that as we go through this, you'll see that there are some suggestions that we have. Again, we -- we leave, you know, those particular recommendations to the relevant workgroups and the Advisory Committee as a whole, but --

Next slide, please. Fantastic, it's working. It's like a miracle. What I'd like to do is just tease apart long-term follow-up a little bit, because long-term follow-up encompasses so much. And, you know, Jeff was a hundred percent right that at the end of the day, what we want to do is make sure that we're improving health outcomes. And a key part of that is making sure that individuals get the care that they ought to get, but there are, really, a bunch of different components that all contribute

to this notion of long-term follow-up.

And, you know, at the -- at the highest level, the way that we think about it is breaking it into -- to two different domains. So, one is all the care and the related special services, educational services and so forth, that individuals receive after being diagnosed with a condition through newborn screening.

And then, there's another component -- I -- I -- I don't want to call it "secondary," necessarily, but -- but a -- a second component to this, which is program evaluation, and that gets to the issues of quality improvements that we've spoken about throughout the day, as well as research, because, certainly, as Scott brought up, there are always questions around these rare conditions about the best ways to provide care and the comparative effectiveness of different approaches to management, and -- and the only way to capture that is continuing research as individuals are identified through newborn screening. And I'm going to drill into that a

little bit more.

We've spoken a lot about the -- the Cystic Fibrosis Foundation as a model for doing this, but I -- I think it's important, at the outset, to recognize how different they are, how different that foundation is in terms of the level of resources that it has and its ability to certify clinics and collect data prospectively. So, I think that, you know, that -- that's certainly an aspirational model, but -- but, again, they're -- they're so different that I -- I think that we need to think about things in a -- in a more wholistic way.

So, next slide. So, this is where I invoke the four components of long-term follow-up that we've all discussed before, including care coordination through a medical home as in-space treatment continues, quality improvements, and new knowledge discovery. Again, I think that that captures those points that I was trying to make before.

Next slide, please. And it's been ten

years since that manuscript was -- was published describing the four components of long-term follow-up. Again, long-term follow-up is -- is -- you know, it's -- it's complex. It entails a lot, and I don't think that we should go glum that it's been 10 years since this was published, and we're still debating a lot of the same issues, because there has been a lot that -- that's been published and that's been done, and that's what I'm going to be going through in the next little bit.

Next slide, please. So, there's certain key aspects that I just want to plant a seed that I want you to think about as we go through. So, first of all, there's really a wide variety of stakeholders when we talk about long-term follow-up. You know, at -- at the core of things, of course, are patients and their families. There's the public health system, the newborn screening programs for example. There's specialists. There's primary care providers, and -- and I will say, as a primary care provider, I was, like,

thrilled to death that there's so much attention being paid to the role of primary care today. There's payers. There's drug and device manufacturers. And then, there's all the other services and individuals that patients and families intersect with over the lifespan. So, I spoke about education, but, you know, there -- there're a million different services that -- that families interact with, and, of course, it depends a lot on the particular condition.

And thinking about that, there's also tremendous variation in the conditions. So, there's differences in epidemiology treatment and timing. As Scott Grosse mentioned before, congenital hypothyroidism is the most common condition that's identified on newborn screening and really lives outside of the specialist world. Oftentimes, the primary care providers often take care of children with congenital hypothyroidism. Then, you get to the metabolic conditions and the hematologic conditions, so forth, and I won't -- won't, obviously, name all of them, but they all

have unique aspects.

And one of the things that makes the epidemiology challenging, of course, is, conditions are -- are increasingly being considered that have late-onset presentation. And -- and so, again, it makes it hard to think about long-term follow-up while, you know, we're waiting -- you know, while it takes a long time for the condition itself to manifest itself.

There's variations in the accessibility of experts and -- and where they are. There's variations in cost, and there's variations in fundamental knowledge about the condition, as well as, just, knowledge more generally among the care providers and so forth. I think it's important to consider issues related to funding for long-term follow-up, and, again, we're going to be digging into that.

And then, there's also the issue of authorization, which I hadn't really thought about before digging into this, but this especially ties back to public health. And for

those of you who primarily operate within public health, it'd be interesting to hear what you say in a little bit regarding authorization and -- and, you know -- you know, where your boundaries are or what it is that you're expected to do.

Next slide. So, this is what I want you to keep in mind, as well, as we go through. So, we all know where we want to go, right, but it can be complicated to figuring out how to get there, and, often, the root is indirect and requires multiple lines. So, based on what we've found in terms of what's out there around long-term follow-up, there's going to -- I -- I would posit that there's going to be no -- no simple way to get there, and we need to think about how to combine these things. I was very proud of this analogy. I, like, came up with it in the middle of the night. So, anyway, I -- I hope it's helpful. If not, I -- I'm -- I still like it.

Next slide. So, the -- you know, I -- I will say, too, it's hard to do these long

presentations on these webinars without seeing anyone, so I hope that -- I hope that if -- if I'm not making any sense, or people want me to expand on things, somebody will jump in. The -- oh, here comes a voice. No? Maybe not.

So, the -- our objective was to conduct a landscape review to inform the Advisory Committee about opportunities for improving long-term follow-up. Again, it's not our job to necessarily come up with specific recommendations, but I do think that some recommendations will naturally come up through this.

Next slide, please. So, the Advisory Committee has already done a lot of work around long-term follow-up. There was a publication from 2011 that reviewed, what questions should long-term follow-up be able to answer and looked at things at the patient-and-family level, the medical-care and the medical-research level, and then at the state and national level, and I would refer the -- refer you back to that particular

document to see all the questions that were developed.

And the -- there are examples of issues that were raised from the Long-Term Follow-Up Subcommittee, as well, that we see borne out in the literature. So, the issue of standardized terminology -- (coughing) excuse me. So, as Scott Shone mentioned, it's incumbent upon us, in the beginning, to be able to track individuals going forward, and one of the barriers to that has been having standardized terminology that can be used in -- in datasets to understand what it is about the individual that -- that we're tracking. Jeff Brosco and Chris Kus spoke about quality metrics, and then another issue, which hasn't come up beyond the -- the brief discussion of the federated model, was issues of information exchange. But that has been something that the Advisory Committee as a whole has been thinking about.

Next slide, please. So, for the purposes of this talk, again, we're focusing on activities

related to newborn screening. So, we're not talking, necessarily, about follow-up of individuals with conditions that are on the RUSP but who are detected other ways. Again, we're very interested in issues specifically related to newborn screening. And as best I can, I'm going to try not to be condition specific because, again, I want to try to draw out the generalizable lessons.

Next slide, please. So, I would be remiss without thanking our technical expert panel and -- and -- and our -- who provided all this stakeholder input. I won't read down the names; I'll just leave this slide here for a few more seconds. But what I do want to point out is that we were very fortunate, also, to have a wide array of experts and -- and people who think deeply about long-term follow-up. Separately, we've spoken to a number of individuals, as well, but these were people who served on the initial technical expert panel.

Okay, next slide, please. So, again, I'm

not going to go directly through the report that -- that you all have in -- in the briefing book, but I'm going to highlight things. So, the -- the first thing is, I would point that most newborn screening programs are involved with long-term follow-up, but the extent and nature of it is variable. There are some components of long-term follow-up that -- that are embedded in and not necessarily considered to be long-term follow-up, but I think the -- with the definition we have, really do hit it.

So, some newborn screening programs have specific referral contracts with specialists and are able to get feedback from those groups that they have contracts with regarding whether or not individuals followed up with them. But, again, one of the things that we heard a lot was that the -- that there was this either absence of responsibility or -- or absence of authority that limits what the newborn screening programs can do, specifically around long-term follow-up. You'll not be surprised to note that issues of

time horizon is challenging, and nearly everyone we spoke to -- and -- and, again, this is not surprising -- is that engaging families in this process is critical moving forward.

Next slide, please. So, we did a -- a horizon search around long-term follow-up -- essentially, putting in the -- in the key words for long-term follow-up and newborn screening and then going through those reports and seeing which ones were really associated with long-term follow-up and which weren't. And this is going to build up what, I think, is going to be a nice library for the Advisory Committee to consider. K.K. and I went through these articles and realized that there was a natural grouping of them, and let me just go through and describe them.

So, the first grouping -- and this is not a hierarchical order, obviously, in terms of importance or anything, but it's just -- just a list of the categories that we had. So, the -- the first type is around recommendations about

how to conduct long-term follow-up. So, that would be things like the reports that the Advisory Committee developed around what questions should it be able to address or other documents that have been developed around how to data share for long-term follow-up. So, those aren't necessarily reports of outcomes of long-term follow-up but provide good information about how to do it.

The second is related to prospective studies of data collected by newborn screening programs for specific purposes of long-term follow-up. The third is for prospective studies of the data collected outside of newborn screening programs for reasons other than long-term follow-up. So, again, two different kinds of prospective data, the second one on that list there, again, being data collected by the programs specifically for this purpose of prospective long-term follow-up and then the other is prospective data collected by other groups. And then, the fourth is retrospective

studies of existing data collected for research or for non-research purposes, so any type of retrospective study.

Now, what I would say from having looked at it is, the retrospective studies are -- are -- have provided a lot of very important information, and it's not surprising. So, retrospective studies are somewhat more feasible because the data are already there, but they have the challenge of being able to link the data to these retrospective studies are often based on complicated linkage between, for example, newborn screening program data and different claims data, sometimes even including things like vital statistics data -- birth certificates, death certificates, or educational data, that kind of thing.

Again, the -- the challenge of these retrospective studies are identifying the datasets and figuring out how to link them together. As with any other retrospective study, either the -- the quality is only as good as the

-- the kind of data that you have. So, sometimes you might be limited to only claims data instead of having, you know, for example, specific patient-level data or -- or laboratory data. But these play a really important role in terms of the evidence that's out there around long-term outcomes.

In terms of -- I -- I just want to highlight one other thing, too. The prospective studies of data collected outside of newborn screening programs, an example of that would be the registries that have been set up for following individuals who are getting specific therapy.

So, for example, there's registries for Pompe disease. We looked at those before, when we were doing the systematic evidence review around Pompe disease. Oftentimes, families opt into these registries, and in that case, Genzyme, which, I guess, is now Sanofi Genzyme, managed the dataset.

So, those kinds of registries can be very

helpful, but, again, you know, the -- the challenge is getting access and making sure that one understands what's in it. But I -- I hope that this classification helps, and I hope that the other thing that it emphasizes is that there's more than one way to get to the kinds of answers that we're interested in.

Next slide, please. So, I'm going to go back and talk about the -- the -- the studies in the first group, the conduct of long-term follow-up. So, these have answered the questions of how it should function. There was a study that was done from the Southeast Regional Collaborative that developed what was called the Business Process Analysis that showed all the parties that were involved and how they would participate in exchanging data. There have been -- studies have been published around data requirements for registries.

Then, there's specific recommendations for making long-term follow-up more feasible. So, that -- that includes things like how to link

results to other datasets. And there was a report that grew out of the Advisory Committee about linking blood spot collection device serial numbers -- so, the -- the serial number on the -- on the filter card -- to birth certificate to facilitate long-term follow-up, as well as short-term follow-up, really. There's work that's been done at the regional collaborative level describing approaches to long-term follow-up.

And then, separate to all this, there have been a number of surveys that were done -- these surveys, now, are -- they were between, like, five and ten years old -- that were done to assess, from the newborn screening program level, what sort of data collection they do, what kind of follow-up activities they're involved with, and what the barriers are to care. So, it's not at the individual level, but at least those kinds of reports describe what the infrastructure, at least back then, was for doing long-term follow-up.

Next slide, please. So, in terms of

looking at that -- that -- that body of -- those papers, I -- I -- I think it's fair to say that long-term follow-up is -- is well defined in terms of, at least people know what -- what they want to do. It still doesn't necessarily mean it's happening, but, again, that people are interested in doing it, that there are models for long-term follow-up. It's been a focus of the regional collaboratives.

One of the things that became clear as we were reading the -- the published literature, as well as the -- the gray literature, is that some good ideas have come down, but funding and sustainability has been a problem. And then, in terms of looking at the descriptions of quality metrics that have been described, it's not surprising, but there's attention between condition-specific quality measures versus more general measures that could be used across conditions.

Next slide, please. So, in terms of the prospective studies in newborn screening, there

are a wide range of different conditions that have been studied across multiple different countries. This -- this work, again, though, is hard to do.

One advantage from these prospective studies is that they include a comparison population. Again, some of the observational studies, especially the retrospective observational studies, you need to be careful about whether or not there's a comparison group or not, because you can easily make the wrong conclusions about the effectiveness of a therapy within a comparison group. In terms of the prospective studies, the -- the longest we saw things go out was six years, and most of these studies were focused on predictors of long-term outcomes based on initial presentation or -- or therapy that was given.

Next slide, please. So, in terms of lessons, I mean, you know, it -- it is reassuring that the data can be collected by newborn screening programs for long-term follow-up to

evaluate specific hypotheses, but when you look at the -- the papers that have been done, you know, obviously, they weren't thinking that, many years later, I would be looking at their papers, trying to figure out what made these things feasible or not, but, oftentimes, they don't specifically address, when they are successful, how they managed to pull it off or the cost that it takes to collect prospective data. And I think that that's a -- an important thing to consider.

Next slide, please. So, let's look at the prospective studies of data that was collected outside of newborn screening. So, just to give you a flavor of the kinds of things: There was a study that was done in the United Kingdom on hearing screening that got captured in our search, but we know that there are other disease-specific registries that are being used to collect long-term data. And so, I spoke about the Pompe disease registry, so I won't repeat that, and then I also mentioned the cystic

fibrosis registry, so I won't repeat the discussion there.

Next slide, please. So, in terms of prospective studies of data that was collected outside of newborn screening, the lessons learned is that, you know, that they're, oftentimes, not done. And given the fact that those registry studies weren't captured in our -- our initial search makes us -- makes me wonder, you know, how -- how well we're leveraging them for prospective research. Of course, registries can also be used for retrospective research.

Next slide, please. So, I spoke in the beginning about the role of retrospective studies, and let me just loop back around to that again and say that they generally fell into two flavors. There's chart audits that are done within specific treatment centers, so that can be helpful in terms of learning about the patients that were seen in that one center, but you can lose a comparator group.

And then, I mentioned before about the

data linkage studies. Those are, oftentimes, more powerful in that you can collect more subjects and do more comparisons, and -- and there's the ability to -- to -- depending on how it's done, to have a -- a comparator group, but, again, there are methodologic challenges associated with doing those.

Next slide, please. So, retrospective studies -- You know, I can't put down that -- that these are efficient for rare disorders. You know, one of the -- the challenges is, you're waiting for the outcome to occur. I mean, it's true, whether or not you're doing a prospective or a retrospective study that you need to have the outcome of interest occur before you can say anything.

Linkage across the various -- various datasets can be difficult, and it -- it -- it's easy to do a linkage study wrong. So, I -- I think that if we're going to be consumers of this kind of work, we need to make sure that things are done correctly. Everyone knows, the claims

data can be incomplete or inaccurate.

One of the things that, sort of, bubbled through as I was reading these different papers, whether they be prospective or retrospective, is that there's this gap, I think, around the association between intermediate measures and long-term outcomes. So, let me drill into that a bit so -- so it makes sense.

So, as clinicians, we're interested in length of life, quality of life, opportunities for children as they become adults, those sort of patient-centered, meaningful outcomes. But those can take a long time to develop, and they can be difficult to measure. So, more often, you may find intermediate measures, so changes in a biomarker or changes in the receipt of some service being given, that -- that kind of thing.

And I think it would help the field if there were better linkages between these intermediate measures and long-term outcomes, from a -- a research perspective, given how complicated it is to do this work. So, these are

rare conditions with outcomes that may not happen for quite a long time. So, I think that that's a -- a fertile area for potential research.

Next slide, please. So, I -- I'm going to highlight some examples of long-term follow-up activities. This is part of our gray literature, unpublished search, as well. This is not meant to be complete. I just want to illustrate some things. I'm sure that I'm going to leave out everyone's favorite long-term follow-up activity, and, hopefully, during the Q&A period, you -- you can bring it up. Again, this is just meant to -- to give the taste of -- of what's out there.

Next slide, please. So, I want to highlight something done in California, where they have a web-based screening information system, which can facilitate referral tracking and coordination. That was the kind of thing that I talked about before. And these centers can provide follow-up through five years of age using an annual patient summary, so, you know, a high-level accounting of how individuals are

doing. There's a Colorado program that includes a legislative requirement mandating reporting of birth defects and other newborn disorders that facilitates linkage of datasets, so vital statistics, hospitalizations, and that kind of thing.

Next slide, please. Can you go to the next slide? There we go. Illinois has a -- an annual report based on data collected for children through 15 years of age. Minnesota has a dedicated long-term follow-up advisory -- it was actually somebody in the technical expert panel -- but they're engaged in a wide variety of tracking of -- of outcomes, and they're also collecting parent-reported developmental status on children.

Next slide, please. Clearly, long-term follow-up activities, in the past, has been a focus of the regional centers, as well as the National Coordinating Center. In the brief time that I have today, I can't go through all these particular activities, but -- but, clearly, data

collection, evaluation, quality improvement have all been a focus.

There's the NBS Connect program, which developed out of the Southeast Region. It includes a patient registry and portal, and it's focused on inherited metabolic disorders.

Sue Berry, before, mentioned some of the work that -- that's going on with the Newborn Screening and Translational Research Network, and in specifically, Longitudinal Pediatric Data Resource, which is a data repository for specific conditions, including, for example, SMA, and it's built on a REDCap database. If we have time, Sue, if you want to comment further on -- on that, it -- that would be great.

NewSTEPs, as everyone knows, and especially from the presentation this morning, has really been focused on the issues of data reporting infrastructure but, thus far, has been focused on short-term quality indicators as highlighted by the timeliness presentation before. Obviously, short-term follow-up is an

important component to the whole process of making sure that newborn screening works, but you know, thus far, most of their work has been around the -- the -- on the short-term side of things. I invite them to comment on -- on future directions that they plan.

Next slide, please. So, let me -- let me just end by summarizing that I think it's important to think of long-term follow-up as having these two different aims: the -- the part around assuring care delivery, but also the part around program evaluation and research. I think there's tremendous opportunities in terms of standardizing long-term follow-up outcome, measures focusing on the quality metrics, thinking about things at the -- the three levels -- the patient, the population, the system level -- expanding use of registries, and expanding support for observational research, whether -- I -- I have retrospective written here, but, obviously, prospective is also important. But I think that -- that there's lots of opportunity

around the retrospective side of things in terms of identifying datasets, linkages, thinking about risks of bias assessment in -- in what these things mean.

So, let me just end by saying, it's impressive to see how much work is going on around the topic of long-term follow-up, but also say that I think that there's tremendous opportunity to continue to think about what the Advisory Committee can do to foster more complete long-term follow-up, with the ultimate goal of improving the health of individuals identified through newborn screening.

So, let me just stop there and open things up for questions or comments.

DR. JOSEPH BOCCHINI: Alex, thank you. That was a really -- a nice presentation, and I think, given where the Long-Term Follow-Up and Treatment Workgroup is, I think it -- it really helps inform their work, in fact, within -- overall, the work of the Committee. So, thank you.

First, we have Sue Berry.

DR. SUSAN A. BERRY: I'm sorry to -- This is Sue Berry. I'm sorry to have the -- the jumpy finger there, you know, with my little hand raising, but I -- Alex, thank you for the tremendous amount of effort that this took. It's a really -- a complicated set of questions and -- and so much at the heart of what we want to succeed at in newborn screening. So, I appreciate all of the attention that you and K.K. played to this.

Couple comments just to add some additional thoughts in terms of resources of extant activities -- I'm going to point out the hard work that NORD has been doing with patient advocacy groups to encourage patient-centered development of data collection regarding specific disorders. That, I think, will be a really valuable resource and presents a really critical element in our plans for long-term follow-up, which is -- is, how the families think we're doing.

The other big resource that we don't always think of is that there are selected rare disease networks, the NCATS, RDCRCs, that have some crossover with newborn screening because they follow disorders that are newborn screened. And the one I specifically mentioned is the very specific issue of -- of urea cycle disorders.

I wanted to bring up two specific challenges that you highlighted that I want to really emphasize. The first one is the idea of the single identifier. We've addressed this issue a number of times through the years since I've been watching the Committee, and there's always this sense that people are afraid to have a single, government identifier. It kind of freaks people out that the carrier from birth, and -- and when people have tried to assign such numbers previously, it's been met with resistance. I don't know if that will still be true, but it's something, I think, we need to be cognizant of as -- as a potential issue.

And the other thing I'm going to

highlight is the issue of sustainability. You mentioned it, but I -- I can't emphasize that enough. I think we've brought it up in a number of these discussions today already, which is that the ability to collect and maintain these kinds of data are -- it's expensive and -- and people intensive, and -- and I just hope, as we consider this, we'll also consider manpower issues, or peoplepower issues, perhaps. Thanks.

DR. JOSEPH BOCCHINI: Thank you.

Bob Ostrander?

DR. ROBERT OSTRANDER: Yeah, hi, thanks. So, that was a great talk, Alex. It's Bob Ostrander, American Academy of Family Physicians. Just want to kind of tell you what my takeaway, 30,000-foot-view impression was after listening to all this, and, you know, I hope it reinforces what we think needs to be done and that there is a tremendous amount of work being done on this, obviously, across the landscape. And the struggles that you pointed out at the end, because we aren't really meeting our goals,

suggests that it's a -- a lot of energy that isn't channeled to produce as much result as it could.

And the other thing that immediately comes to my mind when I hear about all this work that's being done is, as Sue pointed out earlier, and others, if we were to federate this and ask the centers to do it, it would take a lot of commitment and resources. What work that's being done right now is taking resources. I mean, that work isn't being done for free, without time and treasure. And I think we're spending a -- a lot of time and treasure with returns but not nearly the return -- the bang for the buck and the bang for the time we could get if it was more focused.

So, I -- I -- so, my -- my takeaway is two things. One is, I think it is imperative that we, you know, really try to work to use planning to redesign the system, so that all this great work reaps the maximum benefit and, number two, I think we need to look at the resources that we're accessing right now to do some of this

work and see if we couldn't transfer some of those resources to the work that we propose instead of having to generate new revenue streams, and that might make it a little less impossible to achieve a vision over time.

DR. JOSEPH BOCCHINI: Thank you. Next, I have Melissa Parisi and then Chris Kus.

Melissa?

DR. MELISSA PARISI: Hi, this is Melissa Parisi, and I just wanted to follow up some of the comments that Sue Berry made a few moments ago. And, also, thank you, Alex, for your excellent presentation. You certainly raised a lot of the issues related to long-term follow-up.

At the NIH, we, of course, are very interested in research into developing treatments that will improve the lives of people who have newborn screening conditions, and the challenge has been, for us, that many of the studies that really address long-term follow-up, or what we would call natural history studies, are very time and labor and money intensive. But there are a

few mechanisms that have been developed that I think have been reasonably successful at allowing investigative teams to really -- you know, really pursue some of these natural history studies for some of these rare conditions that we're talking about that are the consequence of newborn screening, and I just wanted to highlight a few of those for people on the call.

The first one, of course, was mentioned by Alex and by Sue, the Newborn Screening Translational Research Network, which is, really, a contract funded to the American College of Medial Genetics and Genomics to try to capture the datasets that are so valuable for many of the conditions that are screened in newborns. And this can include conditions that are already on the RUSP or conditions that impact newborns but could have the potential to be added to the Recommended Panel.

And at this time, we are linking the datasets -- some of those datasets to global unique identifiers because, from the NIH

perspective, any data that are generated should, really, be shared as broadly as possible, while, of course, protecting the individual identities of those impacted newborns. And one of the ways to do so is through an algorithm that allows for these global identifiers to be generated and to be linked to that individual, without actually revealing the individual, personal, identifying information. So, that's one of the resources that -- that exists that is funded by NIH.

A second is one raised by Sue Berry, which is the Rare Disease Clinical Research Network, and these currently are -- now there are 22 different RDCRCs or consortia that are studying over a hundred different rare diseases, and a number of these, at least 50% of these conditions that are part of the Rare Disease Clinical Research Network funded -- led by the Office of Rare Diseases Research -- over 50% of these impact pediatric patients, and quite a few have implications in -- for newborn screening. So, I know that the current RFA is on the

streets, and applications are due in October for the next round of these awards, and -- and we hope that some of them will actually include newborn screening-related conditions.

And then, finally, we have a program announcement with specified review for natural history studies for conditions that are either part of newborn screening or could be screened for a newborn. And this is a specific mechanism that invites natural history-type studies, which really can inform long-term follow-up for some of these rare neonatal conditions. Because there's a specified review, the panels are sympathetic to the fact that natural history studies, oftentimes, aren't hypothesis driven. So, there's actually value in having somewhat more exploratory aims. And we've funded a number of projects under this program announcement that have been quite important for the field, and -- and we are about to renew this for another three years. So, that's one of the things on the street.

And then, my final comment is, really, just one about the value of historical controls in terms of developing treatment trials for some of the conditions in newborn screening. I don't want to speak for FDA, but I know that for some conditions, and I think Pompe disease is one of them, having the data where historical controls and -- and, sort of, historical data about natural history actually was really instrumental in being -- in allowing the FDA to approve the intervention or the drug for that particular condition. And so, I think that the value of doing research in this environment cannot be underestimated.

And this also creates the evidence base for adding conditions to the -- the Recommended Panel, but there is one caveat: By virtue of doing newborn screening, we change the natural history of many of these conditions. So, there's always the challenge that historical controls and the historical data about natural history can be problematic in terms of looking forward. So, we

always have to be nimble about trying to gather the new data that inform treatment trials.

That's it. I just wanted to make those comments. Thank you.

DR. JOSEPH BOCCHINI: Thank you, Melissa.

And, next, I have two Committee members: Cindy Powell and Jeff Brosco.

Cindy first.

DR. CYNTHIA M. POWELL: Hi, this is Cynthia Powell. I just wanted to mention the importance but, unfortunately, the lack of evidence-based treatment and management guidelines. I know that efforts have been made, you know, in recent years, by the ACMG and the Genetic Metabolic Dietitians International Group. These are very time-consuming and expensive efforts, and especially for new conditions on the RUSP or -- or being added to the RUSP, but also for, even, conditions that have been on the RUSP for a long time. I -- I think there's a -- a lack of these, and if we're going to really make inroads in terms of quality improvement, these

are, you know, extremely important. Thank you.

DR. JOSEPH BOCCHINI: Jeff Brosco?

DR. JEFFREY P. BROSCO: Yeah, this is Jeff Brosco. So, I -- I have a question for -- for you, Alex, and maybe K.K., and this is a great presentation, and -- and it makes sense of a lot of complex literature. And -- and obviously, a lot of your focus was on published literature because that's what's available, which often leads to -- to research and longitudinal studies.

But if we think in -- for a minute about, you know, in any one state or territory, if we wanted to know whether a child who is eight years old, who was diagnosed with PKU through newborn screening, was getting treatment and appropriate follow-up, and someone was checking in to how he's doing, I guess some states might be doing something, some registries might be doing something, but, in some sense, we -- we don't really have a good handle on what's happening anywhere.

And I just wonder -- the question for you, really, is, you gave us, sort of, the -- the 30,000-foot view in your presentation. Will your report have more detail about what's happening, or at least references, just, say, if we wanted to know what was happening in -- in different places around the states?

DR. ALEX KEMPER: Yeah, so we -- the report'll have more detail, and we'll have references to what happens within individual states where we could find that from either a published thing or -- or looking in the gray literature. So, there's a lot of states where we just couldn't find anything from looking, and we -- One of the restrictions from this kind of work is that we can't do a -- a survey of each state newborn screening program. Certainly, we can reach out to the -- the NCC and see if they have any information for the states that we don't have.

One of the things that I think is -- is interesting again to -- to think about is this

issue of authority, as well, in terms of, you know, what -- what states are allowed to do. So, you know, I've presented some models of -- of some states, like California, that -- that has an active program where they can see who was followed up at one of the sites that they contract with, but, remember, too, that -- that not everyone is -- is followed by a specialty center, right? So, I'm thinking about congenital hypothyroidism, for example.

So, this is a very long-winded, roundabout way to say, we'll -- we'll give you more granular detail on everything that we could find, but, just by nature of the way that we can collect data, it's going to be incomplete.

DR. JOSEPH BOCCHINI: Okay. K.K., did you want to add something? K.K. Lam?

DR. K.K. LAM: Yes, hi, this is K.K. Lam. I work with -- Oh, I'm echoing really bad.

DR. ALEX KEMPER: I -- I would say, we are a great team, and I really should -- I meant to thank K.K., again, for all the work that she's

done to make this possible. So, let me just slip that in right there.

(Laughter)

DR. K.K. LAM: Well, and right back at you, Alex. So, it's a team, for sure. And I -- I don't know if you can hear the echo. I can hear my own echo on this, but --

I just wanted to add, in response to Jeff's comments, that, yes, and in part -- So, the -- the illustrative cases that -- that Alex mentioned -- you know, for instance, California, a bit on Colorado, Illinois, and some of the states that we've identified that -- that do have systems in place, long-term follow-up systems of some sort, right, where they're tracking information at an overall level, which is, I think, what you're talking about -- As we had talked about before in our -- in our discussions, you know, we -- we are going to try and highlight some of the -- as much as we can, the procedures and details about those in terms of, you know, what about this has worked, kind of as models,

basically, a practice that may now help -- may help inform, right, any roadmap that might go on, without fully recommending, but --

And, similarly, in the report, we've highlighted the -- Colorado's system, as I understand it -- now, I know that it always changes, and so I'm still verifying the -- the -- the currency, but the -- part of their system is a -- or, at least, part of their follow-up system and public health surveillance system follows birth defects and at least some of the newborn screening disorders. So, it's -- it's not a newborn screening long-term follow-up, right, but it overlaps. And that's, at least in, you know, a few -- as of a few years ago, had some nice crossover in a model for Waybets (phonetic) that's united these, or at least, kind of, forced, by mandate, these different systems to coordinate.

So, to that degree, our hope is that we will know -- in the -- the final, final report, we will have some of those -- those more -- some

of those particular states of where there's clearly systems set up and ongoing follow-up, where we can try and highlight some of the things that seem to be effective in what they might have to offer and then, potentially, any limitations on a national level, because they're clearly state specific.

DR. JOSEPH BOCCHINI: Thank you, K.K. So, we only have time for two more comments, so we'll go in line to Sue Berry and then Mike Watson.

Sue?

DR. SUSAN A. BERRY: So, I'll keep it quick. This is Sue Berry. I -- I want to -- and help me with NewSTEPs, experts, but I want to say that's something they may know. They may not know what states are doing, but they may know if they are doing newborn screening follow-up, at least just a -- a "yes" or "no," because there are flat out some states that can't and don't. And I -- I -- I think one attribute may be to find ways to at least ask if they -- if they are

doing it. I don't -- and -- and for those that aren't, get some sense of what the barriers are, and authorization is the key there, I would suspect. Thanks.

DR. JOSEPH BOCCHINI: Thank you. Mike?

DR. MICHAEL S. WATSON: Yeah, thanks, Joe. Mike Watson. So, I -- to build on what Sue said, I think we also have to understand why states do long-term follow-up or at least why we think long-term follow-up is important to be done. We already work with -- I mean, we've had meetings of 15- to 17 states and are working with a lot of them right now developing long-term follow-up, but the state programs have more of an interest in public-health kind of issues -- Are people getting the care? Are they seeing specialists, or are they seeing primary care? -- while the provider side is much more interested in -- in the outcomes and, you know, knowing how and certainly in the earlier -- earliest stages of our pilots, understanding, what are the experiences of the other providers, because

that's when we start to find out lots of things we didn't know when we went into the pilot studies.

I would also add that the literature is going to be a little deceptive as to what the current lay of the land is. I think the regional collaboratives are gone now. They're now the Regional Genetics Networks, and -- and I'm still the head of the National Coordinating Center for them, but their focus has shifted over to access of populations -- really, underserved populations -- to services much more than they're being involved in long-term follow-up.

During that transition and the development of the NBSTRN, most of the issues related to long-term follow-up have fallen over to the NBSTRN side, where we now have somewhere between 7,000 and 7,500 babies identified in newborn screening, with any of a large number of conditions, who are being followed at interval visits. Some have finished and have acquired data over several years, and others are

continuing.

You know, we're -- we -- we're running four pilots right now. We'll probably have six multi-state pilots going by next year. We build the clinical data elements and underlying data collection tools for those before we start the pilot. So, that is very much in place. It's where we're not involved in the pilots that they tend not to go with, you know, sort of, goals of compatible data over the long term.

And then, to build on something that -- well, actually, I think when we're thinking about genetic diseases, you have to remember that these are rare diseases. They have a fair bit of variable expressivity, and the underlying genetic etiology is enormously variable. So, it's not a question that can be answered, generally, in a very short period of time.

As to the outcome, there's enough variation that contributes stage of onset and severity that I -- I think it's actually a -- you know, a post-market-surveillance kind of problem,

much like the Orphan Drug Act was developed to address, where, you know, there's a certain amount of data that tells you, you -- you know enough and probably should start screening, but then you put in place, essentially, a -- a -- a requirement for data sharing, much like post-market surveillance does for the Orphan Drug Act, and then you learn, over time -- because with these -- with the amount of variation in genetic disease, finding one doesn't tell you a whole lot about all of them. So, you do need a long-term plan for everybody to be able to improve in the care they deliver to these -- to these babies.

Some things can't be done retrospectively. We can't do -- generally, we can't do much about age of onset and penetrance retrospectively, because we have very biased data from people who are sick, very little asymptomatic-people data that we can use to predict likely outcomes. So, you know, it's a very different kind of dataset in newborn screening.

And then, lastly, I would say, we need to build -- I think Debbie Freedenberg commented earlier that we need to look, when we're dealing with educational delivery, at new formats by which education is delivered, much of it web based, much of it electronic, light bulbs under EMRs and that kind of thing. We're already working with the eMERGE Network and the EHR Workgroups and ClinGen to facilitate interoperability of a lot of this kind of information, but, ultimately, if we can't get the right kind of information we need into EMRs that can then be taken at the time you need to do an analysis, then we're never going to be able to sustain this effort.

So, I think we've got to look at the whole system. The paper we're writing should be done in a couple of months and does look carefully at the nature of the systems in which we're trying to operate these programs. And unless we take a high-level look at -- at -- at the way we approach these kinds of things, I

don't think we're going to do any better than short -- very short-term solutions. That's all.

DR. JOSEPH BOCCHINI: Thanks, Mike. I -- I think your work is certainly going to help inform the work of the -- well, the Workgroup, as well as the Committee.

So, Jeff, as Chair of the Long-Term Follow-Up Workgroup, do you want the last comment?

DR. JEFFREY P. BROSCO: Sure. I'll just say, very quickly, this has been an extremely rich discussion, and I've been taking notes furiously that we'll -- we'll follow up as a workgroup. And then, part of it's going to be, I think -- and -- and -- and Alex and everyone else has really helped separate this out -- you know, what is the follow-up that you need to do for research purposes to show how you improve treatment, what needs to be done for program evaluation to make sure programs are running well, and what, kind of, needs to be done at a population-health level to make sure that every

child with a condition is getting good treatment? And -- and that might be one of the ways we start dividing up how we assure long-term follow-up, is -- is to -- is to separate out those, sort of, three different activities.

So, thank you, everyone. This has been incredibly helpful for moving our workgroup ahead, and we hope to bring the recommendations to the November meeting.

DR. JOSEPH BOCCHINI: All right. Thank you very much.

Let's put up the next slide set, and, Alex, you are back in the hot seat.

DR. ALEX KEMPER: Fantastic. So, I am -- in the interest of time, actually, I'm going to go through this presentation quickly, so that there's time for questions. And let me -- let me just echo what Jeff just said. I -- I thought that conversation was really interesting, and I -- I learned a lot from the -- the whole process. So -- so, thank you for the -- thank you for that.

So, the second project that K.K. and I have been working on is developing this newborn screening technology compendium. Dr. Bocchini talked about it before, but the whole idea of this is to provide, essentially, high-level background information around screening methods diagnostic approaches, and treatment, the idea being that not only the Advisory Committee but -- but -- but, really, the -- the general public is having to -- you know, or -- or interested parties, are having to communicate and make decisions around newborn screening.

And with how fast the technology is changing and with how nuanced everything is, it's important that everybody operate from a -- from the same place in terms of when they're talking about the various screening methods, diagnostic approaches, or treatment. So -- and -- and like I said, everyone knows that -- that things are -- are changing very rapidly.

So, what we're doing is working on developing, you know, sort of, this living

document. I think of it as, kind of, like, the Wikipedia of -- of newborn screening, writ -- writ large. And we're doing this by -- we -- we've, like we normally do, convened a technical expert panel. Looking at the literature, we've identified areas that we think would be helpful for everyone. I am pleased to say that -- that I had a very helpful conversation this morning with Dr. Carla Cuthbert at the CDC, who is also going to be helping us around describing the different screening technologies.

So, we have developed a -- a standard template for moving forward in terms of describing what the particular technology is, what its application is related to newborn screening, issues of, if it's a screening test, screening accuracy versus other outcomes, and that's all -- you know, you can classify it under benefits. And then, we have a harms and risks category related to potential unintended outcomes, false positives, that kind of thing, costs, and that -- that -- resources needed,

where we can find that -- special considerations around regulatory issues, FDA approval or that kind of thing, and key references.

So, let me just, in the -- just -- just to help, I'm going to advance a few slides. Can you go to the next slide? And the next slide? I'm just trying to make sure we get out by 3:00. I don't want to get any -- get in trouble. Next slide. Next slide. Next slide. And, again, if you look in the -- if you look in the briefing book, we have a -- a -- a list of some of the issues that we're going to be addressing, although it's -- it's growing from there, and you'll see that in the next report. This is a list of the general template that -- that I'd spoken about before.

Next slide. And I'm not sure how well this broadcasts on your screen, but this just gives you an -- a -- a -- a sense of what the template will look like. And, again, we'll be cross-linking to the various technology reports, you know, as -- as they populate in our little --

again, I'm thinking of it as, like, a Wikipedia kind of thing.

So, let me -- Joe, maybe I'll go ahead and -- and stop here to see if anybody has any particular questions about where this particular train is going or if they have any advice about what we're doing.

DR. JOSEPH BOCCHINI: All right. This is open for questions, comments. Thank you, Alex.

Mei Baker.

DR. MEI BAKER: Okay. Yeah, just quick, I think it's a wonderful thing to do. One of the things I think that will become so helpful, and I do see your template here, reduce the cost. This is just so hard for me to grasp. Reduce the cost compared with what? So, if you can say, this specific technology, it costs how much -- just putting in there. So, that, I think, will be more useful.

DR. ALEX KEMPER: Yeah, I -- we will try to put in costs where we can find it. What -- what I've learned from starting to look at things

is that cost data are hard to come by in -- in figuring out what's actually included in the cost. The good news is, I'm going to ask -- this -- this may be good news for me but not good news for him -- ask Scott Grosse, who's our resident expert on this kind of thing, to weigh in on things.

I should have also mentioned, when we were doing the Long-Term Follow-Up report, that -- that Scott's been -- was very helpful in that process, as well. But getting back to the issue of cost -- To the degree that we can find it and to the degree that it's valid and reliable, we'll put it in.

DR. JOSEPH BOCCHINI: Thank you.

DR. CARLA CUTHBERT: This is Carla --

DR. JOSEPH BOCCHINI: Yes, go ahead.

DR. CARLA CUTHBERT: I know that time is of issue here, but I just wanted to reiterate that APHL and CDC do provide week-long, hands-on training courses for various technologies in newborn screening. So, we will be very happy to

leverage our experience and the information that we have from those courses to be able to provide information that would be relevant to the -- to the -- the general public, I guess, but to the Committee, specifically, as it -- as it relates to the technology and the screening technology.

DR. JOSEPH BOCCHINI: Thank you, Carla.

Are there additional questions, comments?

(No audible response)

DR. JOSEPH BOCCHINI: All right. Well, we look forward to your continuing work in this area, Alex, and the fleshed-out document. So, thank you.

DR. ALEX KEMPER: Thank you.

DR. JOSEPH BOCCHINI: Are there any -- is there any new business that any of the Committee members wish to bring forward at the present time?

(No audible response)

DR. JOSEPH BOCCHINI: Hearing none, I want to thank everybody for their participation today. I think we've had a really good meeting,

and there's -- from the input that we've had, I want to thank the organizational representatives, as well as everybody who works on the individual workgroups, because it's clear that a significant amount of effort is being made to move projects forward and to -- to bring information to the Committee to inform its work.

So, in summary, many of you are going to be hearing from the Committee soon about either serving on the Steering Committee or being a participant in the upcoming review of our nomination and evidence review processes, as well as our decision-making process, as well, and -- and then, I think, based on the conversations today, we do have a number of items that we will be thinking about how to schedule, going forward, to be on subsequent meetings to then move those topics forward.

I want to remind everybody that the next meeting will be in -- in Rockville on November 1st and 2nd, and that'll be in person, as well as webcast.

So, again, I want to thank everybody for their participation today and thank HRSA for doing all the homework to make this happen. We apologize for the issues related to the videos, but we are going to make them all available to all of the participants, so that they can be viewed in their entirety at your home. And then, we'll look forward to seeing you all in November.

So, thank you. That'll conclude the meeting.

(Whereupon, the above-entitled matter was concluded at 3:00 p.m.)