The Advisory Committee on Heritable Disorders in Newborns and Children

Virtual Meeting

10:01 a.m.

Thursday, February 11, 2021

Attended Via Webinar

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Reported by Garrett Lorm

Committee Members In Attendance

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Jennifer M. Kwon, MD, MPH, FAAN

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C O N T E N T S

P R O C E E D I N G S

WELCOME, ROLL CALL, OPENING REMARKS, COMMITTEE BUSINESS

CYNTHIA POWELL: Good morning, everyone. Welcome to the first meeting in 2021 of the Advisory Committee on Heritable Disorders in Newborns and Children. I'm Dr. Cynthia Powell, committee chair.

Before we begin with committee business, I need to take roll. Starting with committee members, Kamila Mistry.

KAMILA MISTRY: Here.

CYNTHIA POWELL: Mei Baker.

MEI BAKER: Here.

CYNTHIA POWELL: Jeff Brosco.

JEFF BROSCO: Here.

CYNTHIA POWELL: Kyle Brothers.

KYLE BROTHERS: Here.

CYNTHIA POWELL: Jane DeLuca.

JANE DELUCA: Here.

CYNTHIA POWELL: Carla Cuthbert.

CARLA CUTHBERT: Here.

CYNTHIA POWELL: Kellie Kelm.

KELLIE KELM: Here.

CYNTHIA POWELL: Michael Warren.

MICHAEL WARREN: Here.

CYNTHIA POWELL: Shawn McCandless.

SHAWN MCCANDLESS: Here.

CYNTHIA POWELL: Melissa Parisi.

MELISSA PARISI: Here.

CYNTHIA POWELL: And I'm here, Cynthia Powell. Annamarie Saarinen.

ANNAMARIE SAARINEN: Here.

CYNTHIA POWELL: Scott Shone.

SCOTT SHONE: Here.

CYNTHIA POWELL: And going on to our organizational representatives, Robert Ostrander. Debra Freedenberg. Maximilian Muenke.

MAXIMILIAN MUENKE: I'm here.

CYNTHIA POWELL: Steven Ralston. Jed Miller.

JED MILLER: Here.

CYNTHIA POWELL: Susan Tanksley.

SUSAN TANKSLEY: Here.

CYNTHIA POWELL: Chris Kus.

CHRISTOPHER KUS: Here.

CYNTHIA POWELL: Shakira Henderson.

SHAKIRA HENDERSON: Good morning, here.

CYNTHIA POWELL: Jennifer Kwon. Jacob Hogue.

JACOB HOGUE: I'm here.

CYNTHIA POWELL: Natasha Bonhomme.

NATASHA BONHOMME: Here.

CYNTHIA POWELL: Siobhan Dolan.

SIOBHAN DOLAN: Here.

CYNTHIA POWELL: Cate Walsh Vockley.

CATE WALSH VOCKLEY: I'm here.

CYNTHIA POWELL: Georgianne Arnold.

GEORGIANNE ARNOLD: Here.

CYNTHIA POWELL: Thank you. I'm now going to turn it over to Mia Morrison, our designated federal official.

MIA MORRISON: Thank you, Dr. Powell. LRG, can you advance the slides, please? Okay, while they are getting the slides set up, I'm just going to go over some standard reminders that I

have for the committee. I want to remind members that as a committee, we are advisory to the Secretary of Health and Human Services, not the Congress. For anyone associated with the committee or due to your membership on the committee, if you receive inquiries, please let Dr. Powell and I know prior to committing to an interview. LRG can advance to the next slide. Next, thank you.

I also must remind committee members that you must recuse yourself from participation in all particular matters likely to affect the financial interest 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 first received a waiver from HHS authorizing them to participate.

When a vote is scheduled or an activity is proposed and you have a question about a potential conflict of interest, please notify me immediately. Next slide.

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/or announced at the opening of the meeting. For this main meeting or for this particular meeting in the Federal Register notice, we said that there would be a period for public comment. Only with advanced approval of the chair or DFO, public participants may question committee members or others present. Public participants may submit written statements. Also, public participants should be advised the committee members are given copies of all written statements submitted to the public, and we do state this in the RFN as well as the registration website.

All written comments are part of the official meeting record and are shared with committee members. Any further public participation will be solely at the discretion of the chair and the DFO.

And if there are no questions, I'll turn it over to Dr. Powell.

CYNTHIA POWELL: Thank you, Mia. Can we have the next slide? Thank you.

In December 2020, HRSA received the resubmission of the nomination package for MPS II Hunter Syndrome. HRSA has completed the initial review for completeness and the Nomination and Prioritization Workgroup is currently reviewing the evidence submitted in the package. I will continue to keep the committee updated and informed about next steps.

At the December 2020 meeting, the committee voted to approve the Review of Newborn Screening Implementation for Spinal Muscular Atrophy final report. After SMA was added to the RUSP in 2018, the committee developed the report in response to the former secretary's request for a report, "describing the status of implementing newborn screening for MSA and clinical outcomes of early treatment including any potential harms for infants diagnosed with SMA."

After the December meeting, I submitted the report to Secretary Azar and will let the

committee know when we receive a response from the Department of Health and Human Services. The report is now available on the Advisory Committee's website. Next slide.

As you know, the committee has undertaken a review of its evidence review and decision-making processes. In February 2019, the committee initiated this project by convening an Expert Advisory Panel to consider the key components of the review.

In 2020, the committee continued to gather feedback from both committee members and organizational representatives on ways to strengthen the evidence review process including an examination of its newborn screening decision-making criteria and the decision matrix.

This morning, we will address two additional components -- the nomination process and review of conditions already on the RUSP. In terms of next steps, within the coming weeks, I will form a small working group to analyze the information we have gathered and summarize changes

in subsequent meetings.

The purpose of analyzing the entire review process is to improve the process so there will be changes that may impact the information requested in the nomination form and considerations during the evidence review.

I would like to note that any conditions nominated in calendar year 2021 will adhere to the current condition nomination and evidence review processes. We are targeting the beginning of calendar year 2022 for nominators to use the updated condition nomination form and have packages reviewed using updated methodologies. The committee will be kept apprised of this timeline and any changes.

As part of this review, the committee will develop a manual of procedures and consumer-friendly guidance materials to educate newborn screening stakeholders on committee processes. Once changes are finalized, the committee website will be updated with guidance materials and a summary of the updated processes. Next slide.

Committee members and organizational representatives received a draft of the December 2020 meeting summary to review. We received updates to the newborn screening decision-making and matrix discussion. Committee members received the revised draft in the briefing book. Are there any other additions or corrections before the committee votes? Hearing none, do I have a motion for approving the minutes?

SCOTT SHONE: Scott Shone, and I move to approve the minutes.

CYNTHIA POWELL: Is there a second?

JEFF BROSCO: Jeff Brosco. I second.

CYNTHIA POWELL: And all in favor of approval of the minutes, if you could raise your hand using the raise hand function. Has everyone had a chance to indicate their approval on the committee? Anyone opposed? All right. The minutes from the December 2020 meeting are approved. Next slide.

Please note that we have a slight change in our meeting schedule today. The committee will

reconvene from break at 12:35 p.m. eastern time instead of 12:40, and public comments will go from 12:35 to 1:00 p.m. Next slide.

The meeting topics for today, Thursday, February 11th, are as follows. The committee will hear a series of two presentations from Dr. Alex Kemper. The first will be on processes for the review of conditions on the RUSP and the second will be to explore potential updates to the condition nomination form. Next, I will perform a brief update on the committee initiative to develop consumer-friendly guidance materials on the condition nomination and evidence review processes. As noted, in the afternoon, we'll return from break at 12:35 eastern time instead of 12:40, and that's in order to hear from everyone who submitted requests to make public comments. We received these from nine individuals. Brittany Hernandez from the Muscular Dystrophy Association will update the committee on the Newborn Screening Saves Lives Reauthorization Act. Dr. Don Bailey from RTI International will discuss the Newborn

Screening Modernization Project. Mike Hu will provide a statement on his experiences as a parent of two children living with MPS II. Dylan Simon from the EveryLife Foundation for Rare Diseases will discuss the organization's federal and state newborn screening efforts. After Mr. Simon, we will hear from Alyssa Seager, founder of the ALD Alliance. Then, we'll hear from Debra Green from the Sickle Cell Disease Foundation, Niki Armstrong from Parent Project, Muscular Dystrophy, will discuss their ongoing Duchenne Newborn Screening pilot. Our last public comment of the meeting will be from Kimberly Tuminello and Heidi Walls from the Association for Creatine Deficiencies providing an update on guanidino acetate methyltransferase or GAMF deficiency newborn screening.

For the last session of the day, we will have a panel of four presenters discuss Continuity of Operations Planning within the Context of COVID-19. Please note a slight change in the agenda. This panel will begin at 1:00 p.m. Next

slide.

Today, the meeting will end at 2:25 p.m. After the main meeting, the education in Training, Follow-up, and Treatment and Laboratory Standards and Procedures Workgroups will convene from 2:40 to 3:25 eastern time to discuss processes for the review of conditions on the RUSP and potential updates to the condition nomination form from each of the three workgroup perspectives. For all who are interested in attending a workgroup meeting at the end of the day today, we will provide a link to access the Zoom meetings in the chat box. We will also review instructions for accessing the Zoom links this afternoon. Next slide.

Tomorrow, the committee will reconvene at 10:00 a.m. eastern time. We will begin with updates from the three workgroups followed by committee discussion on the workgroup suggestions. After a short break, we'll conclude the meeting with a panel exploring innovations in long term follow-up.

I will now turn it back over to Mia

Morrison, who will provide guidance for participating on the webinar.

MIA MORRISON: Thank you, Dr. Powell. Next slide, please.

Members of the public, audio will come through your computer speakers. So, please make sure to have your computer speakers turned on. If you can't access audio through your computer, you may dial into the meeting using the telephone number in the E-mail with your Zoom link. This meeting will not have an all-attendee chat feature. But we do have a period of public comment scheduled for later today.

Committee members and organization representatives, your audio will also come through your computer speakers, and you will be able to speak using your computer microphone. If you can't access the audio or microphone through your computer, you may also use your telephone to dial in and you may find that number in the E-mail with your Zoom link.

Please remember to speak clearly and

state your first and last name to ensure proper recording for the committee transcript and minutes. The chair will call on committee members first and then organizational representatives.

In order to better facilitate the discussion, committee members and organizational representatives should use the raise hand feature when you would like to make comments or ask questions. Simply click on the icon at the bottom of your screen to raise your hand. Please note that depending on your device or operating system, the raise hand feature may be in a different location. To troubleshoot, please consult the webinar instructions page in your briefing book.

I'll now turn it back over to Dr. Powell.

CYNTHIA POWELL: Thank you, Mia. As I noted in my opening remarks, the committee is currently exploring potential ways to strengthen its newborn screening decision-making processes. Today, Dr. Kemper will begin by delivering a presentation on considerations and approaches for systematically reviewing conditions on the RUSP

for the purposes of gaining additional information about the condition including lessons learned on adoption and implementation at both the state and national levels. After his presentation, we'll open the floor for committee members' and organizational representatives' questions and comments. Before we begin, I would like to introduce Dr. Kemper.

Dr. Kemper is the Division Chief of the Ambulatory Pediatrics at Nationwide Children's Hospital and Professor of Pediatrics at the Ohio State University College of Medicine. He completed his pediatric residency training at Duke University followed by combined fellowship training in health services research and medical informatics with residency training in preventive medicine at the University of North Carolina. Dr. Kemper served as a member of the US Preventive Services Task Force from 2014 through 2018. In 2011, Dr. Kemper joined the Executive Editorial Board of Pediatrics and developed a new section for the Journal focusing on quality improvement.

In 2013, he was Appointed Deputy editor of Pediatrics. I'll now turn it over to Dr. Kemper.

PROCESSES FOR THE REVIEW OF CONDITIONS ON THE RECOMMENDED UNIFORM SCREENING PANEL - RUSP

ALEX KEMPER: Thank you very much, Dr. Powell, and good morning everyone. What I'd like to do in this presentation is just preen the issues around the opportunities that would present themselves for reviewing conditions that are in the RUSP and it's my goal to leave plenty of time for questions and comments, at least as best as we can do via Zoom. Next slide, please.

So, as I go through the presentation, I want you to keep this picture in mind. So, the way I think about reviewing conditions on the RUSP is that when we first review the conditions, there's a lot that we don't know. Things are blurry, there -- there are things that are often uncertain, and this is, you know, caused, as you all know, but a combination of both the rarity of many of the conditions and the fact that issues related to screening and treatment are often time

relatively new, and the advisory committee does a remarkable job of making recommendations in the face of that uncertainly in looking at what's known about the condition and the potential benefits and harms from the whole newborn screening process.

I really see looking at conditions that have been added to the RUSP as having clarity and really better understanding as illustrated on the right side of this picture. Next slide, please.

I put this slide up just to remind you of where this presentation fits into the process that we've undergone in terms of looking at how we can strengthen the review process, and I'll just leave this up here for a couple of seconds and I'm going to share this slide again later. But here we are talking about issues of evaluating conditions that are on the RUSP, and later this morning, I'll be talking about the nomination process. Next slide, please.

So, again here today, we're going to be talking about review of conditions that are --

that have already been added to the RUSP, and this follows along behind a number of other conversations that we've had including the values compensation, the decision-making process, and so on. Again, I just want to make sure that everybody understands the context of the way this falls. Next slide, please.

So, the rationale for looking at conditions that have already been added to the RUSP is it allows us to look at updates in the evidence of screening and treatment for those conditions that are on the RUSP both the primary and the secondary conditions.

Reevaluation of these conditions would allow us to look at new treatments, things that have emerged since the condition was first added to the RUSP to look at new clinical recommendations including issues of clinical management, better understanding of the conditions during infancy. As members of the advisory committee know, we often times struggle with developing things like the case definition and

determining what the primary goal of screening is versus the secondary goal. And over time, as more clinical experience is accrued, it allows you to really better understand exactly what the target of screening is.

It would allow us to look at longer term follow-up. Certainly, long term follow-up both delivery and understanding the outcomes are central to maximizing the benefits of newborn screening. And when we do the initial reviews, the periods of follow-up that are available are often quite short, you know, on the order of a year or two. Looking at conditions that have already been added to the RUSP would allow us to better understand the impact that it has had on public health and clinical services including the availability of clinical services and really look at the impact of expanding newborn screening for the conditions on children and families.

And then, I list at the bottom this issue of being able to look at unresolved issues during previous reviews. So, there has not been an

evaluation where important questions haven't been identified, and this really gives us a mechanism to be able to look back at any unresolved issues and, you know, this is one of the reasons when we looked at the SMA again recently, as presented at the last meeting, that whole report is really focused on open questions at the end of the previous review where SMA was added to the RUSP. I think that's a good model for how that could work. Next slide, please.

So, you know, the issues about this are first of all, thinking about the frequency of which conditions would be reevaluated. So, one could imagine that the advisory committee might just say let's look at conditions that have been added to the RUSP with regular frequency, you know, three years, five years, ten years. I mean, obviously, I just put those up as examples. And, you know, one of the advantages of just having a regular cycle for conditions that have been added to the RUSP is that it would bring to the core conditions that maybe people hadn't recognized as

being important.

Another approach would be just an ad hoc reevaluation of conditions based on changes related to the science or the implementation of screening, new treatments, and those kinds of things. But if there's going to be an ad hoc approach, there needs to be a formalized method to allow conditions to be nominated for review.

Of course, the advisory committee might want to hybrid, so regular reviews unless, you know, something of particular note got brought back up.

Now, I think the principles and the review criteria should be essentially the same for whether or not it's a new evaluation or a reevaluation. But obviously, not all the key questions need to be looked at. So, in partnership with the advisory committee, the specific key questions for the review could be focused and one of the advantages of looking at something that's already been added to the RUSP is this ability to add increased emphasis on things

like implementation or other open questions that were identified before. But the mechanisms -- the approach would be the same as with a new review, just really targeting the key questions of the things that are appropriate and also the ability to look at long term outcomes or linkage to clinical services and those kinds of things that we're just limited in our ability to do with the initial review. Next slide, please.

So, the other issue for the advisory committee is related to what the outcomes of this re-review would be. So, there's an opportunity to clarify the recommendation. For example, the target of screening or whether something should be considered a primary target or a secondary target and that could be tied to advances in screening technology. But, you know, there could also be recommendations about things like short term follow-up involved and long term follow-up.

It would also be an opportunity to inform issues related to newborn screening and care delivery. So, you know, if there were, you know,

specific facilitators to the screening and follow-up process or if there were unexpected barriers, those kinds of lessons learned, it could be helpful for the advisory committee in terms of making new recommendations.

I list removal here. The removal is not the goal. Conditions that have been added to the RUSP have already gone through this fairly rigorous process. So, I just put that out there, but I don't think that that's the primary goal of this re-review process. Next slide, please.

So, this sort of mirrors the picture that I put up before with the art restoration. Again, when the advisory committee makes its recommendations initially, you know, the lay of the land may be a little uncertain. There's often times worry about potential harms or barriers to follow up as would be the dragons and the sea monsters and the reevaluation really gives the opportunity to better understand the geography of newborn screening and if there are unexpected issues or potential harms, things that could be

done to navigate around those. I think I've stretched out that analogy probably as far as I can go, but I hope that puts it into perspective. Next slide, please.

So, I'm going to stop a second and just want to leave questions for the advisory committee. Again, as Dr. Powell mentioned, not everything needs to be resolved right now. But I hope that we're able to have some discussion around these points.

So, first of all, what information would be most important for you to learn about from the re-review? What kind of findings would be helpful in general? In what ways could a re-review help guide improvements to the process of newborn screening and follow-up, especially the issues of long term follow-up? And then, finally in terms of a process thing, I'd be interested to hear members of the advisory committee weighing on whether or not they thing that there ought to be just some sort of, you know, routine periodic reevaluation versus an ad hoc nominated process

based on things that might have changed since the initial evaluation.

And so, now that I've laid out the issues, Dr. Powell, I'll had things back off to you.

CYNTHIA POWELL: Thank you, Dr. Kemper. We'll now take questions and comments from committee members first followed by organizational representatives. As a reminder, please use the raise hand feature, and if anyone has problems with that, just either say something or raise your actual hand. I'll call on your in order of when you raised your hand. Please remember to unmute yourself, speak clearly, and state your first and last name before speaking.

Let's see, and I saw -- Kellie Kelm, did you have a question or comment? I couldn't tell. I saw your hand raised first, but I know you were having some problems raising your hand.

KELLIE KELM: No, maybe I found the raise hand button. But, no, I'm sorry, I don't.

CYNTHIA POWELL: Okay. All right. On my

screen, I'm just going to go to who I see right now. So, Scott Shone.

SCOTT SHONE: Thank you, Dr. Powell. So, Scott Shone from North Carolina. So, I -- I guess, Alex, they took the slide down. I think it might be helpful if we could -- I don't know if that would block Dr. Powell's view of who is in there, but I think putting those questions back up is helpful to think back.

But I think -- I'll start with frequency to review because that was the first thing what grabbed my attention was. So, later on this afternoon, I'll be presenting about the National Contingency Plan, and written into legislation is that it has to be reviewed every five years, where typically something like that is reviewed after a major change. I wonder, Alex, if we can thread the needle on both is that there is a frequency with which the committee looks at the whole panel just to see if there's anything of concern, interest, or change, but then also have built into that review some sort of assessment. I don't know

what it is, and you said we don't have to answer that today. But a new therapeutic -- a new technology to screen for the disorder or something else that we learn. So, I think that the answer is somewhere in between both, if possible, because I don't want to have to wait for a time, but also, it would be good to not forget that we need to look at it.

I think in terms of what do we need to know, I think there's a range of technical from the screening side itself, diagnostic, and obviously on the technical side, you know, recently -- or not even recently anymore -- there is a recommendation on SUAC and Tyrosinemia type 1 and then you look at other types of, you know, targets.

Diagnostic -- therapeutic, but also, I always want to bring us back to, you know, the C in ACHDNC is children, and if we're going to be looking at -- looking at these disorders down the road, it might give the committee an opportunity to start making recommendations or looking at sort

of the transition of these kids into childhood and other recommendations associated with that.

So, while we might be looking at and considering on the newborn screening side of these disorders, perhaps this gives us the opportunity to stretch out legs into the mission of the committee beyond the newborn period of newborn screening itself. That's my comment.

KELLIE KELM: Thanks a lot. I'm going to just jump in there, Scott, while I have the floor. I think these are the slides for the questions for the next presentation, not this presentation.

CYNTHIA POWELL: All right. Next, Mei Baker.

MEI BAKER: Hi, Mei Baker, committee member. So, my question and comment is associated with the secondary condition or secondary target. I think we need to be very careful how we phrase that. I do believe when the new condition is nominated, so in our mind, it's screening for a condition. I think it's important to include the information when you use any markers screening

this condition, what else potentially can pick it up. I wouldn't use condition. The reason I can kind of give you some example -- for example, you use TREC for SCID. You may pick up some [indiscernible 34:04] deletion. This is a condition. But if we are using C3 acylcarnitine to identify the PAMMA, but you also may pick up the maternal B12 deficiency situation. So, the infant condition can be a maternal condition.

And also, let's say we use SMN1 Exon 7 -- homozygous Exon 7 deletion to identify -- to screen for SMA. If you do not have SMA2 [indiscernible 34:46] part of the program, you may pick up the infant with a homozygous Exon 7 deletion with SMA2 copy number 67. Theoretically, that could happen, and this kiddo may not end up in treatment.

So, I think before -- ahead of time, when a nomination is going forward and based on the understanding we have how we can list the potential situation, I think is helpful. And I want to emphasize we may use wording carefully

because secondary condition means do you want them or you don't want them, and not everything is a disease state. Thank you.

CYNTHIA POWELL: Thank you. Michael Warren.

MICHAEL WARREN: Thank you. A couple of questions related to implementation that I'd want to think about. One is as we add new conditions incrementally, what are the implications for states as they are adding this to their panels? How are they funding that? How is that impacting resources that are required both from a laboratory standpoint but also the short-term follow-up standpoint on the public health side? I'm always curious, as I've shared with you all before, where there are opportunities for us on the federal public health side to think about how we supports states and what those needs are. Similarly, what are the gaps that states are seeing in terms of follow-up and access to care for conditions?

The second question I would have related to all of those things would be equity. And so,

I'm not just is screening being implemented and what's that process like, but what does that look like across populations and -- and are we doing this in an equitable way, whether that's by race and ethnicity, whether that's by rurality, but important for us to consider that after we look at implementation. Thank you.

CYNTHIA POWELL: Carla Cuthbert.

CARLA CUTHBERT: Hi. This is Carla Cuthbert here. I just want to share some comments that some of our CDC colleagues had when we were viewing some of these comments. So, one of my colleagues pretty much stated, you know, in terms of the information we would want most to have from the review, it was stated we want to be able to identify the gaps and obstacles to truly fulfill the "promise" of newborn screening essentially. So, again, this is something that is more of a wholistic view, which I think that we really understand.

Secondary conditions, I think -- and again, we don't want to be cavalier about this --

should any of them be upgraded to core, and we want to make sure that there is, you know, a rigorous process for that review, not just well, this looks good. That's not the process here. So, we want to make sure that perhaps that could be a question that could be discussed.

I want to concur with Mike Warren. Health equity did come up. That is something that -- that has been part of the conversation for quite some time now. Are certain populations being disproportionately managed? I think that we -- when we were thinking about this, we thought that the process of screening seems to be fairly equitable. However, the, you know, there may be parts that in the process that are less so. So, is there proper confirmatory testing and diagnostic capability available to certain -- to all populations? Continued care -- are there specific groups that are being lost to follow-up. If so, why and how? So, we're really -- that would really be helpful to inform really the impact of our process.

Long term follow-up -- again, we recognize that there are challenges with sustained funding for this. But again, looking at access to care, looking at patients that are lost to follow-up.

As a lab person, you know, I really wanted to -- I'm specifically interested in, you know, some of the state program challenges and successes, and I really want to emphasize successes because I think that in this conversation, we might just be looking for well, what's wrong. I think that we really should also recognize that as we have spent time doing screening for a number of years that the states may have really come up with really helpful efficiencies that are helping to make things better and to streamline their processes and to really recognize what we have done well and really balance that with what are the gaps and what do we need to really fix and change and so on.

So, for the challenges, identifying risks and issues, identifying perhaps ways to strengthen

testing and algorithms, interpretations, and that again, once we get information about outcomes can really truly feed back into the testing algorithms that we've got. So, that really should be used to help inform how we actually do some of the testing as multiplexing, second-tier tests and other ways that we're doing things, have those things been helpful.

So, again, idealistically, I think having some form of checklist that sort of mirrors the nomination prioritization in a package that we submit initially to sort of close that loop so that the things that we're looking at at the beginning are things that we're looking at to sort of see how have we done, have we done some of this fairly well.

Again, what, you know, these things will all help to guide improvements. And again, as a federal entity and a funder, really I'm looking at what -- where do we need to reapply resources to help with the states.

I want to concur with Scott. I really do

think that some form of hybrid in terms of the -- of the review process would be helpful. I think that we need to be -- whatever we do, we need to be very intentional about it so that we're not leaving out some condition that's not terribly exciting to us at that point in time. So, we really do need to have some sort of regularity associated with the review. So, again, every three years or five years -- three years might be a little tough depending on what we're looking at. But, you know, maybe we can just work that in to the data that we're going to be collecting.

You know, I also do think that there might be opportunity to -- I like Scott's idea of prioritizing things that might look urgent now that need to be addressed now, and so, there might be an ad hoc element to this. But I also think that we may also be able to leverage any of the studies being done right at this point -- at that point in time to be able to bring to the table things that are relevant and that are concerning. I know that that was a lot, but I just wanted to

be able to share some of our thoughts. Thank you.

CYNTHIA POWELL: Thank you. Jeff Brosco.

JEFF BROSCO: Yeah, Jeff Brosco, committee member. So, just to remind everyone, this afternoon, the Treatment and Follow-up Workgroup will be dealing with these kinds of questions and then tomorrow, we also have the panel that talks about a lot of the same kind of overlapping things. And so, particularly for Carla and some of the others who have a lot of detailed stuff, if you could send me -- if you're written that out, that would be great because we want to make sure we cover that. Mostly, I'm trying to take notes.

I think the big message I would like folks to hear right now, though, is there are all sorts of things that we would love to know, and the question is, what does the advisory committee -- what's its role in following up on some of the conditions. And, you know, for example, Carla mentioned how important it is to do some of the lab follow-up, and that's an important thing.

Equity is clearly something that we're going to discuss this afternoon and tomorrow. Which of those things fall clearly on the advisory committee and which might be more a state function or professional organization function or a research function or a treatment group function? And I would say that this is probably -- the advisory committee should probably focus most on what we can learn about the process of the RUSP going forward and thinking all the questions we'd like to answer, that's probably the one that's most relevant to us as a group.

CYNTHIA POWELL: Thank you. Shawn McCandless.

SHAWN MCCANDLESS: Thank you, Cindy. I agree with many of the comments that have been made already, and I'll try to be brief, but I have quite a few notes.

It seems to me that the purpose of the review should be to document the goals of adding a particular condition to the newborn screening panel are being met, and that would involve

collecting certain types of data on a national basis, collecting it from all of the states, if possible. I think that that -- those -- the types of data that we would be specifically interested in would be looking for evidence of benefit of the newborn screening program, evidence of harms related to the newborn screening program, evidence of value of the newborn screening program, and that would include trying to have a good handle on sensitivity and specificity for the disorders, false alarm rates, cost, and other aspects that are related to the outcomes. We will be looking for new information that becomes available regarding natural history of the disease or the altered natural history after initiation of therapy, new therapies, unexpected consequences of therapies, and long term outcomes.

We would also want to, I think, have a mechanism for monitoring for sentinel events related to the process of newborn screening, things that we might not have been able to anticipate at the time that came up, but one-off

things that would point us toward taking a deeper dive at the time of this review.

And then finally, I would advocate that the reviews need to be regularly scheduled, but they're likely to be at different intervals, depending on the condition. And I think that if one were to look at PKU, you would want to -- if you were looking for outcome information and other follow-up information, that review you would want to do down the road in five or ten years. For Pompe where most children die at a year or two of life, a 3-year evaluation would be a perfectly appropriate time. So, I would advocate for some flexibility that's determined at the time that the program is initiated, when should the review be done.

And the last thought I had was that I think that the -- the review should be considered an evaluation, not necessarily -- I don't necessarily think we need to apply sort of the decision matrix to every review. But the review should -- should be done to confirm that we're

meeting the goals of adding a condition to the RUSP to being with, and if there is a question, it's raised in that process, then a matrix-based review of whether the recommendation needs to be changed or removed should follow on. That should be a decision that's made at the time. Thank you.

CYNTHIA POWELL: Thank you. Bob Ostrander.

ROBERT OSTRANDER: It took a second to unmute. This is Robert Ostrander, a key organizational rep. I'm going to try to stay focused on what I think is one of the biggest issues we need to talk about with this, and that is the unintended consequences of these public health measures that are based on small studies and pilots initially and specifically harms.

So, I think one of the main purposes of this should be to identify if unanticipated harms or harms out of proportion to benefit are found. So, I think there should be a focus, number one, of how we choose which conditions -- so, maybe we'll have a schedule for some, but I think

anything that as we, you know, periodically look through the list, whatever sort of workgroup does that, the folks in the room should say were there concerns about potential harms when this was added to the RUSP that we kind of decided did not weigh the reasons to add it or are there concerns now about unintended harms that would prompt a review on the ad hoc basis.

I think, you know, this happens. For those of in general medicine, we see this all the time. I mean, we've kind of gone in a big circle, for instance, in adults with PSA screening, you know, once we started screening for prostate cancer, and just like with these newborn screenings, we're suddenly detecting a whole lot more cases, and, of course, the cases we're detecting are the ones that are relatively milder and may not have needed treatment, and there was a lot of, you know, harm outweighing benefit once that got implemented. But I think there is potentially certainly with some of these conditions that have either milder forms or late-

onset forms that we may harm some folks. And that might be one reason to remove something from RUSP if it turns out the screening, as we did with prostate cancer sort for a while, or at least, you know, make sure that those caveats get in there.

So, you know, that really was my comment is we need to realize that early on -- I think early on, you know, be very aware that we're not causing harm outside of benefit because the implementation ratio didn't match the pilot ratio, probably because of increased case detection.

CYNTHIA POWELL: Thank you. Kamila Mistry.

KAMILA MISTRY: Kamila Mistry from AHRQ. Just a quick question. So, I like Jeff's note about, you know, really attaching our ideas around evaluation to the goal of the committee, because I think we can get very broad in terms of what we're doing and trying to really understand the swim lane or what those goals are, I think, can tighten sort of our purview and I think also help to curb some of the discussion or maybe ground some of the

discussion in the workgroups.

Also, I wanted to ask Alex a quick question around -- I think, Alex, you noted that the evaluation or the metrics, potentially as we could call them, would vary or could potentially vary depending on the condition. Is that -- did you -- is that what you said?

ALEX KEMPER: Yeah. I think what I meant was like the particular -- like the range of questions that would need to be asked might vary. So, you know, things you might ask for -- the PKU or the hemoglobinopathies might be different than Pompe disease, you know, again, I'm just picking on those conditions. You know, it's sort of the old guard versus the new guard or, you know, if there were particular issues about the availability of services or, you know, issues of disparities and those kinds of things. I could see where at the time the re-review began, the advisory committee might -- might, you know, charge us to look at particular things or look for the difference of, you know, specific things. So,

instead of like a one-size-fit-all sort of thing is what I think about. Does that make sense?

KAMILA MISTRY: Yeah. I just think there might be, you know, from evaluation perspective, there might be some systematic answer or core element that we sort of focus on and then, I guess, in addition to because I think that allows us to look at things broadly as a program as well in terms of our goals. Thanks.

ALEX KEMPER: Yeah, thank you.

CYNTHIA POWELL: Anyone else who hasn't had an opportunity to give their opinions or ask a question? Oh, let's see, Natasha Bonhomme.

NATASHA BONHOMME: Hello. I'm Natasha Bonhomme with Genetic Alliance. Thank you for this presentation. You know, I really just wanted to echo a lot of what has been said.

First, you know, I think really thinking about the review process and not necessarily the order of what conditions would be re-reviewed, but really making sure that it is -- I don't know if it's a mix or how we would prioritize it, but that

we aren't just focused on the newer conditions. I think there could be a lot learned from the conditions that we've been screening for decades at this point. You know, my mind definitely goes to Sickle Cell Disease and really seeing where are we at with the stats and how has that changed or ebbed and flowed over the years. So, I really -- I think that would be important.

And then, I know that equity has come up a number of times, which is great. But I think really delving deeper and -- and I don't know if this is just -- well, I don't think it's just within the context of this question, but really broadly just what does equity look like in newborn screening and, you know, from a condition perspective but also from a whole process perspective. I think there is still some digging that needs to be done around that. I think often times, we do lean on when thinking about equity, we look at lost to follow-up, which unfortunately, sometimes those conversations go to well, why aren't parents coming back as opposed to saying

what are the either barriers in the system that are leading to some of those inequities. But I think that really taking -- using this as an opportunity to look at where are all those other gaps as Dr. Cuthbert was saying, I think could be a really good opportunity to go a couple of layers deep in newborn screening around equity, which I know is of great importance to this committee. So, thank you.

CYNTHIA POWELL: Thank you. Anyone else? Okay, I don't see anyone else. Thank you all very much for your thoughts and feedback about this important --

MAXIMILIAN MUENKE: Cindy, there was someone -- Debra Freedenberg had her hand up.

CYNTHIA POWELL: Oh, I'm sorry. I didn't see you, Deb. Go ahead, Debra Freedenberg.

DEBRA FREEDENBERG: Thanks. Yeah, I don't have a hand raise, so thank you.

I agree with a lot of what's been said in terms of what we should be thinking about in terms of review, and I think what I really had wanted to

bring up is that we have a lot of new advances on the "legacy conditions," and that as we think about review, we should incorporate the new therapies and technologies to evaluate our efficacy in terms of newborn screening and the conditions because on a lot of these conditions, when they were put on the RUSP, the newer therapies were not available, and I agree wholeheartedly with Natasha that we do need to look at it in terms of systematic failures that are out there and that, you know, not just left to follow-up but, you know, what is it within the system that's preventing that full equity that we all would like to see.

So, I just wanted to make a very brief comment. Sorry, I can't find a raise hand button anywhere in my system. So, sorry about that. Thanks.

CYNTHIA POWELL: Yeah. Yeah, I don't know, everyone's screen is a little different, but on mine, if you just kind of hover your mouse along the lower part of your screen, you'll see

it.

DEBRA FREEDENBERG: Yeah, I tried that.

CYNTHIA POWELL: If not, just shout out. So, anyone else who can't raise their hand on the screen and has -- wants to make a comment?

Okay. Well, thank you all very much. I think, you know, clearly from our discussion at our last meeting and today, this is a very important thing, you know, that we're planning to do, and as, you know, we mentioned at the beginning, we will be looking at this in more detail. The workgroups this afternoon will have a chance to discuss it more, so we'll be able to get feedback from others who we're not able to hear from this morning.

Also, tomorrow, we're going to hear a round table discussion about some of the challenges in doing this and other considerations regarding longitudinal follow-up. I think, you know, certainly people have brought up important things regarding what the -- what information would be most important and, you know, I've heard

from several of you that, you know, thinking about the equity issues about, you know, what is the outcome of adding a condition to the RUSP and thinking about the, you know, specific condition that we're screening for as well as other conditions that we're -- we're going to pick up, looking at, you know some of the conditions on the secondary list and thinking about, you know, perhaps what, you know, whether they should be moved up to the -- to the primary list on the RUSP.

And I think also, you know, not drawing on everything again but, you know, how are states going to do this? You know, it may be easy for states to give information on, you know, the numbers screened and the numbers detected and numbers confirmed but, you know, I have concerns about longer term follow-up and how we're going to be able to get that information as well as how we're going to be able to pay for it.

So, you know, thank you very much. Let's see. Oh, just looking at some messages that I'm

getting from Mia. So, as I said, we're going to be discussing this further, and we'll report back to the committee with next steps for the review of conditions on the RUSP.

All right. Next, we're going to consider ways to strengthen the nomination process by exploring possible updates to the condition nomination form. Dr. Kemper will present on possible revisions to the nomination form including suggestions that have been provided to the committee within the past few years. These suggested revisions were included in the briefing book, and I'll now turn it back over to Dr. Kemper.

CONDITION NOMINATION FORM

ALEX KEMPER: Thank you very much. So, this is -- this presentation is just going to recap conversations we've had before around the nomination process, and I do have some questions to pose to you all at the end -- questions which you might have accidentally seen a second ago. So, I hope that didn't ruin the exciting mystery

at the end. Next slide.

So, the objectives of this part were to inform the advisory committee about ways to strengthen the overall decision-making process and develop future [indiscernible 59:50] again. That's our overarching work that we're doing right now. Next slide.

And again, I just showed you this before. So, improving the nomination process is where we are now, and again, this fits into the overarching work that we're doing around strengthening the review process. Next slide.

And again, we did things before around the review process, and now we're talking about the nomination. Next slide.

So, this is a screen grab of the process for nominating a condition, which includes points to the nomination package and the next steps. Again, this is what's up on the advisory committee website. Can you advance, please?

And so, you all are well aware of the nomination process, but I just want to recap

things a little bit to help motivate the conversation about opportunities to strengthen it.

So, external nominators prepare the nomination package and they submit it to HRSA. HRSA then reviews the nomination package for completeness and works with the nominators if there are any major gaps in that process, and ultimately the package, once completed, goes to the Nomination and Prioritization Workgroup, which looks at issues related to the seriousness of the condition, whether or not there's a clear case definition, the analytic validity of screening, the potential clinical utility around screening, the available treatments, and whether or not there is prospective pilot data. Again, the advisory committee expects there to be some sort of pilot data before recommendation can be made.

The Nomination and Prioritization Workgroup assesses whether or not the requirements are met and whether or not there is likely sufficient evidence to move forward. It then presents this information to the advisory

committee, who then votes about sending it to the Evidence Review Group for a full review.

Once something is handed to the Evidence Review Group, the full evidence review is -- you know, we've discussed at many meetings the scope of that -- that process has to be completed in nine months. So, there's this critical time element to the process. Next slide.

So, the nomination process is really guided both by what is needed to move something along as well as the needs of the Evidence Review Group once something is handed off. So, the more complete something is, the more straightforward the evidence review process is, so, for example, making sure that the case definition -- the target of screening is clear and that kind of thing.

And so, the question has come up about how can the nomination process be more aligned to the evidence review process or even down the line, the decision-making process that the advisory committee would have to go through. So, again, by strengthening the information that comes in, then

the review process and ultimately the decision-making process can be streamlined and made more efficient.

So, the question has come up around, you know, what additional information could be there, and what information is important to have early in the process. And certainly, from reviews we've done like SMA, even though the experts are in the field, as it comes, you know, on the nomination process, it helps us to put together our technical expert panel.

There are a lot of things that could go into the nomination process that would expedite the review process. And, of course, moving forward, if the nomination form is modified, there will have to be work done to make sure that the nomination or the potential nominators and the public are aware of these changes and Dr. Powell talked about certainly if the nominal form and process were to change, that would not be retrospectively applied to the current nominators and put them at any sort of time disadvantage.

Next slide.

So, this table shows how the current nomination form looks and then potential areas of expansion. So, of course, it has the nominator's contact information, but -- and this has been a good starting place for us in terms of identifying who the experts are but explaining that would be helpful. The form provides information about the condition and the treatment, but the more granular that is, the easier that is in the evidence review process. You can see that the form asks about standard metrics regarding screening, but even more detail, if it's available, would be helpful including pilot study contacts, algorithms, and so forth.

The nomination form asks for key references from published articles. But providing even more information on available registries for the condition or if there is important unpublished data would be helpful.

So, you know, of course the tension is the degree to which the nomination form provides

all that information versus the potential work the nominators have to do. But having all the information that's listed on the righthand column would certainly facilitate the review process. Next slide.

So, actually, you know, could we go back to the previous slide for one thing? I neglected to say something. Even though the nomination form, you know, could be expanded to include these features, I don't want anyone to think that the advisory -- that the Evidence Review Group would take shortcuts in terms of not doing all of our usual due diligence in terms of looking for unpublished data, assessing the quality, and that kind of thing. So, again, this is just providing additional information to help point us in directions, but it wouldn't change our usual process in terms of looking for everything that we could possibly find and also assessing the qualities. So, certainly the work that we would do would go beyond this. Next slide, please.

So, in terms of opening this up for the

advisory committee, I'd ask you to reflect on whether or not the elements from the nomination form and what's needed for the evidence review line up, is there anything that's missing. And so, I'd appreciate your thoughts about it.

And then, if the nomination form does change, you know, what are the opportunities that are available to facilitate the nomination process. So, we don't want to make the nomination process burdensome or really, you know, more difficult to do than it is already. But we would like to capture this additional information if it's available, so making sure that we're able to be clear and communicate to our stakeholders and the public is obviously important, but I wonder if there are other opportunities that are out there that we're not thinking about that could facilitate the nomination process.

So, with that, Dr. Powell, I will hand it back to you to facilitate the discussion.

CYNTHIA POWELL: Thank you. Thanks, Dr. Kemper. We'll now take questions and comments

from committee members first, followed by organizational representatives. And once again, a reminder, please use the raise hand feature if you can. I'll call on you in order of when you raised your hand, although it's a little difficult from my screen to be able to tell whose first. Remember to unmute yourself, speak clearly, and state your first and last name before speaking. Okay, I'm just -- Mei Baker.

MEI BAKER: Hi. I think I'm going to try to plea my case one more time and kind of -- I think the nomination form, even now that it's done so, when we screen, I mean, nominate a condition for screening for and we also -- I would suggest asking information about if you use specific markers, what else you may also identify because I think -- I personally feel like a bonus and it doesn't do harm. But also, we should have prepared so the committee may have a harm. So, we need to be aware and when we [indiscernible] a condition, take this into consideration and additionally, an example I want to give to you,

for example, XALD and you identify Zellweger syndrome. In my mind, that's not bad. It's good. And so, it's why I try to avoid the use of the term to define the scenario. I think each scenario has a different scenario. But I think we should phrase the way to ask this kind of situation. Thank you.

CYNTHIA POWELL: Scott Shone.

SCOTT SHONE: Thank you, Dr. Powell. This is Scott Shone. So, I -- Mei, I think that -- so, that is asked, right? So, I guess, Alex, I'm trying to understand some of the tweaks you want to make to the form itself seem to be -- to help facilitate the evidence review, right? Not that we're talking here about changing the N&P process or what we're evaluating as part of N&P to evidence review, and before I quiet down, the reason I ask that is because having been part of the last few N&P reviews, I feel like we have -- at least I have had a good line of what is part of our assessment, what are the questions that the Nomination and Prioritization Workgroup are

looking at to make a decision about them pushing a condition to the floor -- let me back up -- about recommending for the advisory committee to recommend for full evidence review.

And so, I have another question, but I just want to make sure I understood your presentation correctly that that's part of the goal here.

ALEX KEMPER: Yeah. And, you know, certainly, I don't want to step on the, you know, outside of my purview, and so I'll have Dr. Powell comment in a second. But, you know, there's a tension when you -- when the Nomination and Prioritization Workgroup evaluates something because there's going to be uncertainty regarding whether or not it should be added to the RUSP at the time something goes through the Nomination and Prioritization Workgroup. You know, we don't want the Nomination and Prioritization Workgroup to, you know, spend so much time trying to look at everything and then only put those things in where, you know, there's a high level of certainty

that something will be added to the RUSP, because that, you know, the whole point of the evidence review process is to kind of lay there everything that's known about newborn screening. And so, I would hate to push so much stuff to the Nomination and Prioritization Workgroup that they feel like they need to do a complete evaluation before things go through.

The lens that I was taking on it is both to provide additional information that may help you, you know, in terms of, you know, like you're full of certainty before you push things through. Again, are you up to that level that's 100 percent? But also, just reflecting on the fact that we have nine months to put together a high-quality evidence review and the individuals who are nominating the condition are often times, you know, the national and the international experts in the condition. And so, to the degree that they can provide us more information to make sure that we can really come to a good case definition that we really know what's out there and what's not out

there would be really helpful.

So, I don't see this as like a wholesale, you know, disrupting of what the Nomination and Prioritization Workgroup does, which is already a difficult enough task.

Before I ask you whether or not I answered your question, I'll have Dr. Powell weigh in just to make sure that I didn't cross any lines that I'm not supposed to cross.

CYNTHIA POWELL: Yeah. No, I agree. I mean, I think it's not just information for the full evidence-based review but also for the Nomination and Prioritization Workgroup to consider as much information as is available, you know, and certainly published information is a lot easier to get at and review, you know, versus, I know Alex, especially for what you and KK do, you know, trying to dig up the gray data is very challenging and time-consuming. So, you know, we definitely want to work together with the groups that are putting forth nomination packages and I guess we'll be able to talk a little bit about

that in terms of some of the consumer-friendly materials that we hope to develop.

ALEX KEMPER: And just to build on that again, I don't want to make the bar so high for nominators that people are dissuaded from nominating because they feel like they need to have, you know, all this additional stuff that is really to help us know which path to go in. Does that -- Scott, does that answer your question?

SCOTT SHONE: Yeah. No, absolutely it does because I think that what -- I think that what you've added -- what you've recommended to be added to the form makes a lot of sense in that aspect. So, I agree with that. And I don't think it's an -- I don't -- my perception is it wouldn't be a significant burden to request that additional information because I actually think it will inevitably help conditions that are ready to move forward to evidence review, help the N&P group get to that decision.

I do feel that what we ask for now when nominators have the data and complete it

thoroughly is very -- I think it's very well thought out and the group that brought that together prior to me joining, I think should be commended for really how well that process was thought out in advance of evidence review. And I think that I'd like to commend HRSA and the team, who have done a lot of work around educating nominators and helping facilitate package production or giving feedback. We've had a couple recently where we've provided feedback, and I think it's great. So, the minor tweaks, I think this is a great quality improvement review and these minor tweaks that are being recommended, I feel only benefit without adding a substantial burden to the process.

ALEX KEMPER: Thank you.

CYNTHIA POWELL: Thanks. Jeff Brosco.

JEFF BROSCO: I agree with what Scott said. And Alex, I think that the original, you know, Scott and I have been in a bunch of the Nomination and Prioritization Workgroups over the last few years, and I think the process is pretty

good and HRSA has done a great job, and your suggestions are all excellent ones I think can be done.

I think tying this to the previous conversation we just had between 10 and 11:00, I think there's a critical piece we're missing and sort of minor piece that would be nice to add. And that critical piece is if you think about how Carla framed follow-up, "Are we fulfilling the promise of newborn screening?" and Shawn's was really nice too, "Are our goals being met?" I think we should ask the nominating group to say what are the goals and a couple specific measurements. So, is it merely mortality? Is it ventilator-free survival? Is it a quality of life measure? Saying here's how we know we've been successful. They're in the position because there's, you know, there's families, there's advocates, there are the world experts in those conditions, and they're probably in the best position to be able to say here's how we know we've been successful. That helps all the rest of

us sort of judge, okay with a likelihood of getting there and in longer term, seeing whether it's been true or not.

So, I think asking the nominating group to say what are the two or three or four key outcomes measures that we can look at and say yes, we hit a home run or we're not quite there yet would be really useful.

And then sort of a minor second thing is at least say something about the prospect of longitudinal follow-up. You know, we have this registry in place -- you sort of mentioned that -- and here is how we could going forward keep track of whether we've met those goals or not. And I think we've mentioned in previous years before the lost year of 2020, that this wouldn't be part of the nomination scoring, right, because some groups obviously have a lot more resources than others, but at least saying we have a national registry, two-thirds of kids are in it, you know, we think this could keep going forward, and we'd be able to judge whether we met our goals or not.

ALEX KEMPER: That's very helpful. Thank you.

CYNTHIA POWELL: Thank you. Robert Ostrander.

ROBERT OSTRANDER: Yeah, hi. Robert Ostrander, AAFP. I'm going to just -- I thought I might be able to lower my hand when I saw Jeff up there talking about long term follow-up.

But there are two parts of the longitudinal follow-up. Question one of them is the data collection to see if we're meeting our goals and that would be wonderful to have -- to see if there's a plan in place -- if they could put that on the nomination form -- is, you know, like all the forms you fill out online, you know, it's optional or this field is not required. But I do recommend that we have a required field that says what -- not what the data gathering of longitudinal follow-up is -- but actually the care portion of longitudinal follow-up. And we on the Long Term Follow-up and Treatment Subcommittee run into -- we've had this sort of dichotomy

discussion about what do we mean by longitudinal follow-up. Do we mean data gathering and success or do we mean actually the clinical side.

And, you know, as a clinician, I always think about longitudinal follow-up as actually being the care. And I think there should be a requirement that there be some image of what happens after the initial intervention to follow these kids. It doesn't mean the system has to be in place, but I think the word we've used, you know, before our hiatus this year, you know, was some sort of a blueprint or a vision. If there isn't a blueprint or a vision of how that's going to happen -- how these kids are going to be taken care of after the initial intervention, assuming it's a one-and-done intervention, which some are and some aren't, or even, you know, the foods for the metabolic diseases. If there isn't a vision of how -- how that's going to be done, then I don't think that it's ready for addition to the RUSP. I mean, there has to be an idea of how that's going to happen, and I don't think the

pilot studies necessarily provide that, and that's what we're relying on right now for our evidence review.

So, I would like to see a line on there that has to have something in there of what our vision is for the treatment side of longitudinal follow-up as opposed to the data-gathering side of it if that makes sense to people.

CYNTHIA POWELL: Yes, thank you. Jed Miller.

JED MILLER: Yes. Hi, Jed Miller Association of Maternal and Child Health Programs. I think at one point last year, we had discussed the value of knowing the resources behind a given nominator and I know on the first page of the, you know, of the form there. It talks about the nominator and the organization, and then any other sponsoring organizations, and I'm just wondering based on that, you know, would there be value in having a disclosure of sorts to say this is how much, you know, money -- the total operating budget of this organization, for instance, and if

any money has been specifically allocated for this nomination process. And I'm wondering if that would give a better context for the nomination, and thinking about, you know, people who have fewer resources, whether that would help level the playing field, so to say.

And, you know, I might go so far to say one could argue that that top information could be redacted, right, from everybody else so that it's purely on the merits of what, you know, the rest of the package is and everything. I presume that some, you know, you could infer who perhaps nominated something. But I'm just thinking about that, you know, that context and seeing, you know, it seems like there's a lot of value in knowing what's behind, you know, a given nomination. Thank you.

CYNTHIA POWELL: Thank you. Melissa Parisi.

MELISSA PARISI: Hi. This is Melissa Parisi from NICHD/NIH. I just wanted to make a brief comment sort of following up what Mei Baker

was saying earlier and a couple others have commented on, which is the whole issue of the case definition.

I think that when a nomination packet is put forward, that the nominators do their best effort at case definition and, you know, I think that that's a very reasonable starting point, but I wonder if it's worth asking is there the potential for the case definition to be expanded, and by that I mean, you know, as we know from some of the examples given, such as XALD also picking up Zellweger Syndrome, and we know that X-linked SCID ended up being, you know, a much broader case definition.

In some situations, actually, the case definition gets more refined through the whole process of actually reviewing the evidence and then having experience with the condition as in the case of SMA. So, I'm just thinking that if there's some way to capture in maybe a speculative manner from the nominator, ways in which they think. There may need to be some modifications to

the case definition based on the experience that accumulates. That might be a helpful consideration as well because I think that, you know, we do our best when we're trying to anticipate a given condition's screening needs but then there's really nothing like the benefit of having more experience to help us understand a little bit better what exactly is being picked up on.

CYNTHIA POWELL: Thank you. Natasha Bonhomme.

NATASHA BONHOMME: Great. Thank you. Natasha Bonhomme. Two points that I think -- actually, maybe they're one -- and my focus is on this last question in terms of opportunities to facilitate the process, and I think taking this on is really great and having this conversation is really important, but also to note not everything can be embedded in a form. And, you know, a lot comes from -- no matter how hard you try, a lot comes from conversations with other people and, you know, being able to go through and see how the

nominations have happened in the past. And so, I just want to be able to throw that out there. Probably, gosh, it may be almost close to eight or so years ago, Genetic Alliance was tasked as through some other funding to be a technical assistance around the nomination process, where people would just call in and say -- and I triaged a lot of those calls -- and a lot of times, it was like am I on the right path, I saw this group do this, I saw that group do that. And again, I say that just to say there are some things that can't be necessarily captured in a form.

And also, I wanted to comment on, you know, I really liked Jed Miller's idea in terms of other information that we could capture, and again, wanting to say that maybe there are things that we don't need to capture right at the beginning that, you know, these groups don't just magically disappear once they go through the nomination process. You know, there may be some types of data that we want to capture on the back end that will then help inform the next group that

goes around. So, not thinking of it just in terms of that one nomination process, that one nomination of that condition, but as a whole entire system and what can we learn and kind of iterate as we go condition by condition. So, those were the -- the main pieces that I just wanted to share. It doesn't have to be everything in this, even though it's really clear a lot of work and thought has gone into it.

CYNTHIA POWELL: Thank you. Anyone else who hasn't had an opportunity? All right, Annamarie is seconding what Natasha just said. Anyone who is having an issue with the raise hand feature who hasn't been able to comment? I'm not seeing anyone.

So, thank you all. You know, I think again this is also an important area. I've heard, you know, people say in the past like the nomination form is deceptively easy and there is a lot more that needs to go into it, although, you know, being on the other side and wanting to get as much information in a timely manner as

possible, you know, it is challenging, and we do, you know, benefit, I think, from having as much information as possible. I think, you know, overall people feel it sounds like, you know, it's -- we're just looking at some tweaking versus any type of major revision that it works quite well already. Thinking about issues of case definition, which is really something that we can't, you know, is often not available at the time the committee initially, you know, reviews things. But that will be important.

Shawn McCandless, I see you have your hand up.

SHAWN MCCANDLESS: Yeah, thanks, Cyndy. This is Shawn McCandless. You said something that I think triggered something that I've been reflecting about recently, and I'm relatively new to the committee, so I may not really fully grasp the intricacies of the mission and the expectation of this committee. But it does seem to me that the nomination form is not so much -- it's not sort of one group of people asking another group

of people to do something for them. It's really a way of formally initiating a relationship, a process that you're going to enter together with the committee and the nominator to develop a package of information and evidence that's needed to -- for the committee. The committee ultimately has the responsibility to make the decision. The nominators have the responsibility to support the committee and help gather the evidence that's needed as much as they can with lots of resources available through Alex's group and others.

But to me, I think it's really important that we keep in mind that -- that it's not -- it shouldn't be a competitive or it should be viewed very much as a relationship-building opportunity of mechanism, and therefore, it's not sort of a you put it in and then you cross your fingers and wait to see what happens, but rather you put it in and you enter into a relationship and start this process together to collect data and come to a decision.

CYNTHIA POWELL: Cate Walsh Vockley.

CATE WALSH VOCKLEY: Hi, Cate Walsh Vockley, National Society of Genetic Counselors. In the interest of seconding comments, I wanted to second Jeff Brosco's comment about including information about goals of newborn screening, particularly because this gives an avenue or an opportunity to get some of the parents' perspectives since they are involved in the nomination process. We hear very different perspectives from folks like the Newborn Screening Round Table, and I think definition of goals might give us a broader perspective from the parents' side, and I think that's really important. Thanks.

CYNTHIA POWELL: Thank you. Mia Morrison.

MIA MORRISON: Thanks, Dr. Powell. This is Mia Morrison, Designated Federal Official, and I think that this has been mentioned, but I'd just like to reiterate that HRSA and Dr. Powell, we're available to provide technical assistance to groups that are nominating conditions, and I think

that Scott Shone mentioned earlier that we have been providing TA to different nominating groups earlier this year. So, I just wanted to state that.

CYNTHIA POWELL: Thank you. And as with our earlier topic, you know, the workgroups this afternoon will have an opportunity to discuss this and hear from workgroup members who otherwise may not have had an opportunity to give their opinions and provide feedback. And then, we'll hear tomorrow morning from the workgroup chairs or co-chair about that.

And let's see, anyone else? Let me just double check my list here. Okay. All right. Let's see. So, we're running a little bit ahead of time, but that's okay. We can go on. I was just going to give a brief presentation about some of the consumer-friendly materials that we're hoping to develop, and I can see those slides. Thank you.

CONSUMER-FRIENDLY GUIDANCE MATERIALS

CYNTHIA POWELL: All right. So, as part

of this effort working with the group from HRSA and others, hearing all of your feedback, we plan to develop educational resources directed to newborn screening stakeholders on the nomination and evidence review processes. You know, we understand that the nomination form is complex and as Mia just said, you know, getting feedback ahead of time, you know, we think is very helpful and certainly, you know, possible. I think what happens more often is that, you know, the nomination package is submitted and then, you know, HRSA goes over it very carefully and lets people know what -- what information is missing and sometimes, you know, that's a quick fix, sometimes it may be take longer.

So, we will be incorporating changes from review of the evidence review process and hope to have some frequently asked questions or list available on the website, and we welcome, you know, additional feedback from other stakeholders regarding what, you know, might be most helpful in this.

Any -- any additional comments or suggestions about this process? I saw something. Okay. Let me just check. Let's see, Natasha, did -- did you have a comment about this or something? I see your hand up, but it might be from before.

NATASHA BONHOMME: Yeah. I didn't know if you wanted comments on this piece in terms of consumer-friendly materials yet or if you were still going in to add more slides. So, sorry.

CYNTHIA POWELL: No, I don't think we have any more slides. So, go ahead.

NATASHA BONHOMME: Yeah, obviously this is a topic I'm passionate about. But, of course, I'm really excited to see that this effort is underway, though I know it's been something that's been discussed for quite some time. So, I think this will be really great and really value add. I don't know who the right person would be to share this, but, you know, in I believe it was 2016 or '17, Alex, KK, and I had a number of different draft materials really focused on, you know, how do we explain the nomination process. We had some

text and some diagrams. You know, happy to share that. Just let me know who to forward those materials to. But I'm just wanting to say that we're, you know, excited to be able to see this part happen because it's really important to make sure that the consumers and people who are actually going through this process have these types of resources. So, thank you for this effort.

CYNTHIA POWELL: Thank you. Anyone else? Okay. Let's see. Let me just check with Mia. We're running a little ahead, but probably nobody would be too disappointed to have a longer break. Is that what we want to do, Mia?

MIA MORRISON: Yeah. I think we can just give everyone a longer break and we'll still plan on reconvening at 12:35 for public comment.

CYNTHIA POWELL: Sounds good. Thank you.

BREAK

CYNTHIA POWELL: Welcome back, everyone. I'm going to take attendance again. So, I'll call the roll. Kamila Mistry. I think she's going to

be a little delayed. So, she'll be back. She'll be rejoining us shortly. Mei Baker.

MEI BAKER: Here.

CYNTHIA POWELL: Jeff Brosco. Kyle Brothers.

KYLE BROTHERS: Here.

CYNTHIA POWELL: Jane DeLuca.

JANE DELUCA: Here.

CYNTHIA POWELL: Carla Cuthbert. I think she may be a bit delayed getting back also. Kellie Kelm.

KELLIE KELM: Here.

CYNTHIA POWELL: Michael Warren.

MICHAEL WARREN: Here.

CYNTHIA POWELL: Shawn McCandless.

SHAWN MCCANDLESS: Here.

CYNTHIA POWELL: Melissa Parisi.

MELISSA PARISI: Here.

CYNTHIA POWELL: I'm still here. Annamarie Saarinen. I think she was possibly going to be a little late too, so. Scott Shone.

SCOTT SHONE: Here.

CYNTHIA POWELL: Robert Ostrander.

ROBERT OSTRANDER: Here.

CYNTHIA POWELL: Debra Freedenberg.

DEBRA FREEDENBERG: Here.

CYNTHIA POWELL: Max Muenke.

MAXIMILIAN MUENKE:

CYNTHIA POWELL: Steven Ralston.

STEVEN RALSTON: Here.

CYNTHIA POWELL: Jed Miller.

JED MILLER: Here.

CYNTHIA POWELL: Susan Tanksley.

SUSAN TANKSLEY: Here.

CYNTHIA POWELL: Chris Kus.

CHRISTOPHER KUS: Here.

CYNTHIA POWELL: Shakira Henderson.

SHAKIRA HENDERSON: Here.

CYNTHIA POWELL: Jennifer Kwon.

JENNIFER KWON: Here.

CYNTHIA POWELL: Jacob Hogue.

JACOB HOGUE: Here.

CYNTHIA POWELL: Natasha Bonhomme.

NATASHA BONHOMME: Here.

CYNTHIA POWELL: Siobhan Dolan. Cate Walsh Vockley.

CATE WALSH VOCKLEY: Here.

CYNTHIA POWELL: Georgianne Arnold.

GEORGIANNE ARNOLD: Here.

CYNTHIA POWELL: Okay. Anybody who's joined in the last minute or so that didn't get a chance to answer before? Okay, thank you.

So, now, we're going to go onto public comments. In accordance with the Federal Advisory Committee Regulations, members of the public had the opportunity to register to provide written and oral public comments. The committee received one written public comment on newborn screening for WHIM, W-H-I-M Syndrome, an immunodeficiency condition. We received eight requests to provide oral public comment. Please note that three of the individuals who will speak also submitted a written summary of their remarks, which were distributed to the committee.

First, we will hear from Brittany Hernandez, and I'll give you the order of

speakers. Brittany Hernandez, Dr. Donald Bailey, Mike Hu, Dylan Simon, Alyssa Seager, Debra Green, Niki Armstrong, and last Kimberly Tuminello and Heidi Wallace. So, first we'll hear from Brittany Hernandez.

PUBLIC COMMENT

BRITTANY HERNANDEZ: Thank you, Dr. Powell, and thanks to the committee for the time today. I will be brief. I want to provide a short update on the reauthorization status in Congress of the Newborn Screening Saves Lives Act. As everyone here knows, the authorization for the program lapsed in 2019. I have had the pleasure of working with other stakeholders across the newborn screening community towards a positive reauthorization of the program. I am pleased to report that the reauthorization bill has been introduced in the House side by our longstanding champion Lucille Roybal-Allard of New York, and we anticipate quick introduction on the Senate side by the same Senator Maggie Hassan here relatively soon.

I also wanted to share some information provided by March of Dimes regarding funding levels. So, for 2021, both HRSA and the NSQAP got $1 million increases and we are continuing to advocate for 2022 for robust increased funding for the work across HRSA and the work that all of you are doing. If there are any questions from the committee members or anyone else on the call about the status of the Newborn Screening Saves Lives Reauthorization Act, I will be happy to take them, and I am easy to find at the Muscular Dystrophy Association. So, thank you very much.

CYNTHIA POWELL: Thank you very much for the update.

Next, we'll hear from Dr. Don Bailey. Don, are you available?

MIA MORRISON: Dr. Bailey, if you're on the line, please raise your hand.

CYNTHIA POWELL: Here he is. We can't hear you, Don.

MIA MORRISON: LRG, is it possible to unmute Dr. Bailey?

VINCENT: Unfortunately, it's not on our end. We're looking okay over here.

MIA MORRISON: Okay. Thank you, Vincent.

Dr. Powell, why don't we go to the next public comment, and hopefully Dr. Bailey can troubleshoot while we hear from our next commenter.

CYNTHIA POWELL: Okay. We'll hear next from Mike Hu.

MIKE HU: Thank you, Dr. Powell, for the opportunity to speak here. Hello, everyone. My name is Mike Hu. I'm a father of three boys. Ten years ago, my two older sons were diagnosed with MPS2, which I'm sure you are familiar with, as it's RUSP nomination is being reviewed.

In my boys' case, a single point mutation caused the key lysosomal enzyme IDS to completely lose its function and large pseudomolecules accumulate in other bodily tissues leading to organ malfunctions, developmental delay, and will eventually lead to premature death.

Diagnosed around the same time at the age

of 4 and 1, both boys have undergone nearly a decade of standard enzyme replacement therapy, and the younger one has so far fared much better both physically and developmentally. One key difference is his early diagnosis at the asymptomatic stage, thanks to his brother's diagnosis, and the early start of treatment when physical and neurological damages were limited.

Imagine if they were both diagnosed as newborns and treated immediately. Our journey with MPS2 has turned me into a devoted advocate. Three years ago, I resigned from my full-time job as a molecular diagnostic test developer to focus on newborn screening initiatives, which I believe is key to systematically winning the battle against devastating genetic disease like MPS2.

As a parent, I deeply believe in the collective effort and landmark work by the ACHDNC since its inception. As an advocate and a diagnostic industry veteran, I believe there's a lot more we can do to improve the system and help more babies and their families. We're witnessing

the takeoff of exponential growth in technology and therapeutic innovations. What has happened in the past decade was unimaginable ten years ago. To stay ahead of the curve, we must start making systemic changes that can help accommodate disruptive technology advancements and maximize the therapeutic benefits of these innovations.

I appreciate the effort from the committee to have started reviewing the process for improvements. Other comments which we will hopefully hear from Dr. Don Bailey and Ms. Alyssa Seager have some excellent points about the RUSP nomination system. I will not provide any spoilers, but I do want to emphasize how critical it is for us to revamp the system now.

The current newborn screening system was established out of the needs for fundamental changes and has been a textbook example of systemic innovation. It is up to all of us to keep up the spirit, embrace the new challenges, and enact changes to make possible a brighter future when all affected babies will get timely

diagnosis and treatment and lead better lives. Thank you.

CYNTHIA POWELL: Thank you very much. I think we're able to hear Don Bailey now. Don, can you go ahead?

DON BAILEY: Yes. Can you hear me now?

CYNTHIA POWELL: Yes.

DON BAILEY: Great. Good afternoon. Sorry about that. Thank you for the opportunity to speak today. I think I know most of you. I'm Don Bailey, Distinguished Fellow at RTI International and a former member of this committee for six years.

Today, I'm pleased to represent a consortium of rare disease leaders that are collaborating to identify ways that newborn screening can prepare for a likely rapid expansion of transformative treatments including cell and gene therapies in the coming decade.

In September of 2020, the consortium, which includes EveryLife Foundation for Rare Diseases and leading rare disease companies

announced an independent effort to evaluate the capacity of newborn screening in the United States to provide timely diagnosis of all newborns who may benefit from new treatment if and when such treatments are approved for use in the United States.

My team and I at RTI, one of the world's leading nonprofit research institutes will complete the first phase of the project. It's a study of the strengths and limitations of the current system. The study team has convened five panels with an outstanding group of key stakeholders across the newborn screening spectrum to assess the readiness of the system to keep pace with medical innovation and to identify potential solutions that meet the needs of people with rare disease.

So, the study -- we're moving very quickly. The study will be completed about May of this year when preliminary findings will be available and published. The consortium is very pleased, as you can imagine, that the committee

continues to consider recommendations for strengthening your processes and the system at large, and we sure hope that the findings of our modernization assessment will inform and support your efforts.

If the committee wishes, I would be pleased to present a full report of findings and recommendations at a future committee meeting. So, thanks very much for allowing me to speak today.

CYNTHIA POWELL: Thank you. Next, we'll hear from Dylan Simon.

DYLAN SIMON: Thank you, Dr. Powell. I would like to thank the committee for providing the opportunity to address you today. Again, my name is Dylan Simon, and I'm the Newborn Screening Policy Manager at the EveryLife Foundation.

The EveryLife Foundation is a nonprofit, nonpartisan organization dedicated to empower the rare disease patient community to advocate for impactful science legislation and policy that advances the equitable development of

[indiscernible 29:31] diagnosis, treatment, and cures. Today, I want to give you an update on our recent newborn screening initiatives.

At the federal level, as Brittany discussed, we've been part of leading the rare disease community coalition efforts dedicated to the passage of the Newborn Screening Saves Lives Reauthorization Act. We are excited that the House reviewed this legislation on January 25th, beginning the vital process of working to ensure that this important piece of legislation is finally signed into law. Later this month, we are hoping to convene with nearly 800 rare disease community advocates through our Rare Across America event, during which time they will have the opportunity to educate the representatives on the importance of newborn screening and seek their support for legislation.

We will continue to work with the rare disease community to ensure that the impact of passage of this legislation will have on patient communities as well as understood by policy

makers.

We also remain focused on shortening the timeline between when a condition has been added to the RUSP and what is screened for at the state level. As those present are aware, patient advocacy organizations worked for decades with community leaders to develop the evidence necessary to ensure that RUSP nomination packages are ready for review by this committee. Once conditions have met the evidentiary requirements of that condition, those same organizations and patient communities then face an additional hurdle of state implementation, a process that requires significant resources and many more years. The newborns go undetected and lack access to life-saving therapies and innovations.

The EarlyLife Foundation and RUSP legislation ensures that states must screen for all RUSP conditions within a specified amount of time following a condition's addition to the RUSP, while also ensuring that there's a long term planning choice for a newborn screening program to

facilitate this implementation. Within states where this legislation has passed, we have worked with the state labs and legislators to ensure that both resources and timeline requirements meet the needs of each program.

We have already successfully passed legislation in California and Florida and this year we will be pursuing similar legislation in Arizona, Georgia, Ohio, and North Carolina.

Lastly, in December, we convened a panel to review the impact of COVID-19 on newborn screening programs as part of our 2020 workshop. We heard from state labs and state leaders and are in the process of [indiscernible 31:28] to a manuscript. One central theme throughout our discussion was the importance of considering newborn screening as an essential service to ensure that there's a plan in support of the programming times of public health emergency and that the plan must encompass a full range of services rather than just be focused on emergency planning for laboratory services. We look forward

to continuing discussions to identify changes we need to make in emergency preparedness planning and to ensure that the positive lessons that we have learned from COVID can be integrated into our policy moving forward.

I want to thank you again for the opportunity and we are excited for all the great work that's occurring within the newborn screening space, and we're looking forward to working with you to continue how to effectively navigate and engage within the community. Thank you so much.

CYNTHIA POWELL: Thank you. Next, we'll hear from Alyssa Seager. Is Alyssa available? Ms. Seager? I think not. We'll go on to Debra Green if she's available.

ALYSSA SEAGER: Hi, I'm here. Can you hear me?

CYNTHIA POWELL: Yes.

ALYSSA SEAGER: Thank you. So, hi. I'm Alyssa Seager from the ALD Alliance, and I'll be speaking on behalf of the EveryLife Foundation.

Dear Chairwoman Powell and members of the

Advisory Committee for Heritable Disorders in Newborns and Children. On behalf of the over 30 million Americans living with rare diseases, the EveryLife Foundation is pleased to offer the following comments to inform the advisory committee's ongoing conversations about the review process for new RUSP nomination packages.

To inform our policy work, we convene the Community Congress, a forum for collaboration representing over 200 individual rare disease patient advocacy organizations in addition to over 90 other health care and biotechnology organizations. Our Newborn Screening and Diagnostics Working Group is dedicated to ensuring that patients receive their earliest access to lifesaving opportunity. Newborn screening is the first step in reaching a diagnosis, preventing the diagnostic odyssey that impacts too many. It eliminates disparities in accessing a diagnosis, ensuring equitable access to earlier intervention and treatment. This provides individuals with the right to a healthy life, continually proving that

newborn screening is one of the most successful public health programs in the country.

Over the next decade, we anticipate an increase in the number of RUSP nomination submissions. Since the creation of the RUSP, patient organizations have led the nomination effort for multiple conditions, often spending years generating the evidence needed to submit a successful nomination. While efforts continue, the current requirements make it impossible to bring forth RUSP approvals fast enough to keep pace with the opportunities innovation is bringing to our community.

We are aware that the committee is working to evaluate how conditions are reviewed. As you consider recommendations for improvement, we urge you to consider the following significant challenges.

Successful RUSP nominations require prospective population-based pilots that may cost millions and take years to complete, which is not fusible for many patient organizations. Without

newborn screening, it is challenging to satisfy criteria regarding treatment and intervention metrics in the absence of early diagnosis. The same challenges associated with developing a treatment for rare disease will exist when assessing the benefit of newborn screening. Disease rarity, heterogeneity, and other disease-specific considerations may impact the ability to assess the benefit of newborn screening within a population.

Recognizing the significant workload of the committee and the pipeline of conditions that may be nominated, we urge you to consider the following to accelerate the review of new disorders to the RUSP. The assessment of the benefit of screening for new conditions should recognize the challenges above. Accepting a degree of uncertainty regarding the amount of data available following the approval of a treatment or intervention and include other sources such as patient and community insights.

The FDA may approve an indication broader

that the population studies in clinical trials. The committee should consider opportunities to leverage FDA decision-making regarding the benefit of treatment in infants and young children to create a central database to track incoming data for conditions planning to submit a condition to the RUSP. This will provide the ability to better evaluate long term outcomes for RUSP conditions.

We are sincerely grateful for all of the work and dedication to our rare communities. The EveryLife Foundation and our Community Congress Newborn Screening and Diagnostics Working Group are ready to support your work, and we look forward to engaging with you over the next coming months. Thank you.

CYNTHIA POWELL: Thank you. Is Debra Green on? If you could raise your hand to be identified so that we can get you unmuted. No? All right. We'll go on to Niki Armstrong.

NIKI ARMSTRONG: Hello. On behalf of Parent Project Muscular Dystrophy, thank you for the opportunity to speak today. My name is Niki

Armstrong, and I serve as the Newborn Screening Program Manager for PPMD. I am pleased to be presenting here about our Newborn Screening Duchenne Pilot in New York state. For the last seven years, PPMD has been leading a national effort to build a newborn screening infrastructure for Duchenne in the US aimed at developing the evidence to support Duchenne newborn screening. This initiative and the associated collaborations have resulted in multiple publications as well as diagnostic tools and resources for primary care providers and families. Our Duchenne effort has convened experts and established the partnerships required to implement nationwide newborn screening for Duchenne. PPMD's Duchenne Newborn Screening Program incorporates expertise from leaders within NIH, HRSA, SCA, CDC, AAP, the American College of Medical Genetics and Genomics, past CMD pilots, the broader newborn screening community, and the Duchenne community.

As a result of all these collaborative efforts, in October of 2019, we initiated a

Duchenne Newborn Screening Pilot in New York state. The pilot was designed to set up, validate, and conduct a consented pilot screen for infants born at select hospitals in New York state and utilize this tool, resources, and expertise at PPMD, the Newborn Screening Translational Network, and the New York State Department of Health.

Out pilot is being funded through a unique model. PPMD has convened a pre-competitive consortium of biopharmaceutical industry partners with a commitment to early diagnosis and intervention in Duchenne. Consortium members currently include PPMD, PTC Therapeutics, Sarepta Therapeutics, PerkinElmer, Solid Bioscience, and Pfizer, Inc. The pilot is guided by a steering committee comprised of representatives from federal agencies, provider groups, and from key Duchenne's stakeholder communities. The pilot is utilizing the FDA-approved CK-MM assay.

More than 20,000 babies have been screened in the state of New York as of the end of 2020 and three newborn boys with Duchenne Becker

and one female carrier have been identified. Families with a child with Duchenne or Becker are followed in the health system's associated Multidisciplinary Neuromuscular Clinic. Parents complete surveys to provide their input on the family's perspective. The pilot has now surpassed the one-year mark and continues to demonstrate the unwavering commitment of our partners and strength of the infrastructure, even as it proceeded at ground zero of the pandemic.

We are so grateful to the leadership within New York state, within the state laboratories, the birthing centers, the specialty clinics, and the primary care provider sites. The ability of the teams to pivot, to continue to enroll families by changing to remote enrollment, and then to see the families via telehealth showed extraordinary dedication. We are grateful to all those working with us to ensure that the babies identified through this program are receiving the most immediate expert and comprehensive follow-up care possible.

Today, we would like to extend our gratitude to the families, experts, and partners who have helped us get this far. With four approved therapies and a research pipeline filled with potential therapeutic interventions, newborn screening will provide optimal opportunities for care and treatment in Duchenne. Our Duchenne Newborn Screening Pilot in New York state is an exciting and critical next step in improving outcomes for children with Duchenne. Thank you.

CYNTHIA POWELL: Thank you. And finally, we'll hear from Kimberly Tuminello and Heidi Wallace.

HEIDI WALLACE: Hi, good morning. My name is Heidi Wallace, and I'm the parent of two children with GAMT deficiency. My oldest daughter is 17, and she was diagnosed at 5 and suffers from intellectual disability and is not independent. My 9-year-old son was diagnosed at birth, and he is in every way a typical 9-year-old. I'm also the president of the Association for Creatine deficiencies and I work in the Utah Public Health

Lab in the Newborn Screening Informatics Program.

And I'm here today with good news. We have successfully identified a baby with GAMT deficiency in newborn screening in Utah and for just a little history lesson for anyone who is new on the committee, we were -- GAMT was nominated five years ago for consideration and in that very same meeting, was when the new criteria were created prior to the vote. And unfortunately, the one true positive was the criteria that we did not meet, and a lot of committee members kind of felt like their hands were tied by those rules they had just voted to implement. And so, by one vote we did not move forward, and so, we've spent five years hoping for one baby to be identified, and so, we are hopeful to be back in May and have a vote on GAMT.

And just a little more background on GAMT. It can be -- it is detected by elevated guanidinoacetate, which can be multiplexed with existing amino acid and acylcarnitine screening. It does not require an additional punch of blood,

it does not require an additional instrument or laboratory technician. It's frequently referred to as a no-brainer for newborn screening. The treatment is over-the-counter supplements that cost less than $100 for the first year of life. We have children who have grown up diagnosed since birth because of siblings, and so, we have good evidence that this is not, you know, impossible treatment. It's a very successful treatment and we're excited to be back in May.

And I do want to just address that criteria. I know that the committee is considering changing the criteria and I want to, just on behalf of all of the very rare disease groups like ours, tell you the difficulty that that criteria places on us. We are rare. We have a small community. So, we don't have a lot of money. Our treatment is over-the-counter, safe, and effective, but it's not backed by a big pharma company who is paying for pilots and paying for advocacy work and what not. And so, for a rare disease that is such a no-brainer, slam-dunk

disease in every other way, this criteria has been very, very difficult, and unfortunately, we have even come to know families who have been born since five years ago whose children have displayed symptoms and have been diagnosed the hard way.

And so, I would encourage, you know, deep thought about that one criteria. When we came five years ago, there was a very thorough research paper written by Dr. Marcia Pasquale, and many experts have told us there is more data and scientific evidence in that paper -- in that research that Dr. Pasquale did than in finding one child. It, you know, it -- the diagnosis did confirm the levels that we have seen in reserve dried blood spots and retrospectively testing those. So, you know, it -- it reaffirms what we know, but it did not reveal anything new. And so, we're just really hopeful that that's one of the criteria that is being reconsidered so that the committee's hands aren't tied and best decisions can be made for other disorders down the road.

So, we look forward to coming back in May

and thank you for your time.

CYNTHIA POWELL: Thank you. Is Kimberly Tuminello also going to speak? Can you raise your hand if you're on the call. Okay. I think we need to move on. Thank you all very much for your comments today, and we look forward to further discussions.

We're going to move on to our next session on Continuity of Operations Planning or COOP and COVID-19. The pandemic has highlighted emerging needs and COOP planning for state newborn screening programs. For example, most COOP plans do not incorporate strategies to address the prolonged and widespread impact of disruptions caused by the current public health emergency. This panel, comprised of representatives from the Texas, North Carolina, New York, and North Dakota newborn screening programs will highlight their experiences with COOP planning and enactment as well as future considerations.

After we hear from the four panelists presentations, there will be an opportunity for

committee members and organizational representatives to ask questions and engage in discussions. Excuse me.

Before I turn it over to our first speaker, I'd like to introduce today's panel. Dr. Susan Tanksley is the APHL organizational representative and the laboratory operations manager in the laboratory services section of the Texas Department of State Health Services in Austin. She manages the day-to-day operations of Texas Public Health Laboratory, which encompasses the state newborn screening, clinical chemistry, microbiology, environmental chemistry, and emergency preparedness laboratories. Dr. Tanksley received a Ph.D. in genetics from Texas A&M University and has been certified as a high-complexity laboratory director through the American Board of Bioanalysis since 2005. She is also the committee's organizational representative from APHL.

Dr. Scott M. Shone, a current committee member, is the director of the North Carolina

State Laboratory of Public Health. He is a board-certified, high-complexity clinical laboratory director trained in molecular microbiology and immunology. Dr. Shone spent nine years as the director of the newborn screening laboratory for the state of New Jersey. During his tenure, the program expanded screening from 20 to 55 disorders and maintained essential services during multiple states of emergency. Dr. Shone is a member of the Newborn Screening Technical Expert Panel for the Clinical and Laboratory Standards Institute.

Joyal Meyer began her employment with the North Dakota Department of Health in November 2011. She has served as the director of the North Dakota Newborn Screening and Follow-up Program since January of 2015. She has experience working with the maternal and child health population and Optimal Pregnancy Outcome Program and the Coordinated School Health Program. She has worked in both the hospital and clinic setting with the prenatal population and has provided nursing care to mothers and their newborns following their

birth. Joyal's educational experience includes a Bachelor of Science and nursing from MedCenter One College of Nursing in Bismarck, North Dakota and a Master of Science and nursing administration from the University of Mary in Bismarck, North Dakota.

Finally, we'll hear from Dr. Michele Caggana, who is the deputy director of the Division of Genetics, chief of the Laboratory of Human Genetics, and the director of the Newborn Screening Program in New York. She is board certified in clinical molecular genetics by the American Board of Medical Genetics and a fellow of the American College of Medical Genetics and Genomics. She is involved in many newborn screening efforts and works with the CDC and the APHL. She is the chair of the APHL Newborn Screening Committee and a member of the National Advisory Child Health and Human Development Council.

I will now turn it over to Dr. Tanksley.

CONTINUITY OF OPERATIONS PLANNING (COOP) AND COVID-19

SUSAN TANKSLEY: Good afternoon. Can you hear me okay?

CYNTHIA POWELL: Yes.

SUSAN TANKSLEY: Great. I want to thank the committee for allowing the four of us to speak to you today about continuity of operations in newborn screening. The pandemic has really made newborn screening programs even aware of the importance of having a good continuity of operations plan, and so today, next slide, we want to talk to you. First, I'll be giving a background on COOP in newborn screening, some of the challenges that we faced, some that you are probably very aware of, but I'll remind you of them.

I'll be giving some information on the impact of COVID-19 on newborn screening that the Association of Public Health Labs has gathered and some of the resources that have been put together for newborn screening programs. Dr. Scott Shone

will be talking about the Continuity of Operations Plan as well as his experiences in North Carolina. And Joyal Meyer will be giving her perspective on COOP from North Dakota. And then Dr. Michele Caggana will be speaking about her experiences during the pandemic in New York. Next slide.

So, just a reminder of what Continuity of Operations Planning is, and this is from the Newborn Screening Contingency Plan Version 2. So, the COOP for newborn screening program and its public health lab should have two basic features. One being a comprehensive pre-identified list of core testing to support activities -- basically, kind of a list of everything that you need to have in place if you have to rebuild. And then secondly, an actual plan of action to ensure that those core activities can be continued without delay. Next slide.

And I borrowed this slide from Stan Berberich, and when we think about emergencies -- and that's what we're really planning for with Continuity of Operations -- in the context of

newborn screening, it's not just disasters. There are the big things like our current pandemic, like hurricanes that have been faced, but it could be even small things. For instance, we had to implement our COOP because there was an FDA recall on a kit that we were using for second-tier testing, and so, the ability to continue operating to continue testing. And so, an emergency for newborn screening is anything that can prevent timely identification or adequate intervention for babies who are born with the disorders that we are screening for. Next slide, please.

So, I want to take you back to 2005 when Hurricane Katrina hit Louisiana and Iowa was able to provide emergency newborn screening for the residents of Louisiana with those babies. And so, and I borrowed some slides, again from Stan Berberich, and so, I want to thank him for sharing those with us.

So, Hurricane Katrina hit New Orleans on August 29th of 2005. On the 30th is when the levees were breached, and then on the 31st of

August is when the Louisiana Public Health Lab realized that they had to have help. And they instituted the EMAC, which is the Emergency Management -- it's a compact -- and Iowa responded to that on September 1st. During that time frame between September 1st when you look down there, as early as September 7th, Louisiana sent the first batch of specimens. So, it's a very -- it's about a week timeframe, but if you consider the fact that there wasn't an actual agreement between Louisiana and Iowa at that time, the immense amount of work that had to happen during the timeframe. On September 6th is when Iowa was selected and the details were finalized. But the very next day is when Louisiana sent that first batch of specimens to Iowa. Next slide, please.

And so, because there wasn't a plan ahead of time, all of the logistics had to be worked out in that very short timeframe. So, some of the details are these gaps that are listed here. Just, you know, the day-to-day operations, how would specimens get sent to Iowa, which facilities

would they be receiving them from, what do they do if they have missing information or rejected specimens, how are they going to report results?

And then, they had to acknowledge a lot of things and decide how they were going to handle those differences. So, they had -- there were different disorders that the two states were screening for. They had different testing methodologies. There are policies that may differ between states such as babies who have been transfused, when a baby is collected and what that might -- how that impacts an actual test result.

And then, from the perspective of Iowa, how do you rapidly increase your throughput? Next slide, please.

And so, another detail that had to be worked out was the legal construct for the agreement between Louisiana and Iowa, and they chose to use the Emergency Management Assistance Compact, EMAC, and EMAC is a congressionally ratified organization that provides the form and stru7cture for interstate mutual aid. The most

important issues that EMAC resolves is the liability issue and then the reimbursement. So, how will the state that's doing the work for another state actually be paid for that? And that agreement was activated in one day. So, next slide.

So, after the emergency situation is over or at least the emergency state part of it is over, it's always important to have an after-action discussion and note the lessons learned.

And so, these were some of the initial lessons learned that the state of Iowa put together.

First of all, it can be done. Even though there wasn't a plan in place to begin with, they were able to work together and rapidly make those decisions and initiate the testing in Iowa for Louisiana.

There is capacity within the newborn screening community, and EMAC was probably the -- the biggest lesson learned from Hurricane Katrina, and you've probably heard Stan talk about EMAC and

its importance, and he definitely stressed that with newborn screening programs around the nation. Next slide.

So, the partnership that was formed between Iowa and Louisiana was very successful. But it served as a reminder of the urgent need for states to establish COOP plans. And just a reminder that every emergency is going to have its own fingerprint. It, you know, every situation is going to differ to some degree, and although you have a plan, it will require adaptive creativity to implement it. Even though the planning that took place between Iowa and Louisiana was extremely short, it was essential that those decisions be made -- be worked out ahead of time. Next slide.

So, New Jersey had learned a lot from Louisiana's experience with Hurricane Katrina and so, Scott Shone was the director in New Jersey at that time, and he had talked with Stan Berberich about EMAC, and they had -- and New Jersey had started to meet with EMAC about its potential use

in the case of an emergency. The Public Health Lab had been placed organizationally within emergency preparedness and had recently moved onto the campus of the New Jersey State Police, which also housed the Regional Operations Emergency Center. So, there were a lot of things that came together and as Scott had entitled this slide, Preparedness, Luck, and Serendipity, because it was just amazing how these things had come together, not without planning.

NYMAC had also been working on a regional COOP plan as well. And in addition, New Jersey Newborn Screening Program, who was contracting with UPS for courier services, had started having discussions with UPS about COOP, and it just so happened that the representative for UPS at the time was pregnant, and so, she had a very -- she was very engaged in newborn screening.

So, at that time, New Jersey didn't actually have a formal COOP in -- a formal COOP developed, but basically everybody had begun talking about it. Next slide, please.

So, on October 25th of 2012, is when the New Jersey Newborn Screening Program started to realize that Hurricane Sandy just may impact them. And so, at that time, the program started planning what they would do if they had to shut down. And so, they started a lot of key discussions with different partners. You know the message, newborn screening is essential, and they started having discussions about how would they get samples to the lab if the infrastructure that they had set up in the state shut down. Next slide.

So, on October 29th is when the storm started to come onshore and they were pretty much now just not in an impact zone but a direct hit. Because they had had so many discussions with UPS ahead of time, UPS had already decided that they were shut down, but they did make provisions to deliver to the lab that day. So, that day the New Jersey program had 19 staff in the lab, and they managed to wrap up all the testing of all specimens that they had in the lab so that there wouldn't be any samples left over. And they had

to begin planning for alternate specimen delivery options because they had already been told that UPS was shutting down. They were already shut down, but they had made that like last delivery to the program, but that they would have to find alternate delivery if they were going to receive any specimens. And so, they began having discussions with the New Jersey State Police. Next slide, please.

And so, this is a picture of the New Jersey lab where newborn screening occurs, and that's what it looked like on October 30th or I should say prior to Hurricane Sandy. Next slide.

And then this is a shot from inside the lab on October 30th and those are solar panels that are over the atrium, and there was glass everywhere, and the building was on emergency generator power. So, staff went in and assessed the building so they could determine the impact of the storm and whether they would be able to process. Next slide.

At the same -- so, during this timeframe,

you can see the impact of Hurricane Sandy on New Jersey. Next slide.

So, on October 30th also, the Regional Operations Emergency Center was in full activation, and Governor Christie was there along with many other people, and one of those individuals happened to be the Commissioner of Health for New Jersey, which was very fortunate. At that time, Governor Christie approved the use of State Troopers to get specimens to the lab. There was communication out through the New Jersey Hospital Association and that hospitals were to transport specimens to the Regional Medical Coordinating Centers and then the New Jersey State Police transported the specimens to the laboratory at 4:00 that day. There were only 7 staff who could make it into the lab that day, but they worked hard to get all the specimens processed that they could. Not all hospitals were able to get samples to those medical coordinating centers that day, but they did get a lot of specimens that day. Next slide, please.

So, looking at the rest of the week, things started -- the lab took over communication the very next day with the hospitals. Follow-up staff had to move to -- relocate to the laboratory because their building was shut down. And additional staff showed up to work on the 32nd [sic.] UPS resumed delivery except for a few hospitals on November 1st. And then on November 2nd, there were only two hospitals that remained effective and normalcy had returned to the lab. Next slide, please.

Again, after any critical event, it's important to have after-action reports so that you can record lessons learned, and some of those lessons learned are recorded on the right side of the slide there from the newborn screening perspective.

And so, it's just really important to note those things because those are the details that you then take back to your Continuity of Operations Plan and build into it. Next slide, please.

So, this slide is just a reminder to everyone out there that newborn screening is a system. It's not just the newborn screening program. There are so many partners that are part of the process. And the newborn screening program itself can only control one part, and that's the program. This holds true for COOP as well. And so, the newborn screening program can't be the only party within a COOP plan. There has to be communication between and across all of the partners. Next slide, please.

So, if you break down the newborn screening process, you can see where some of those system partners fit in. And so, in the preanalytical aspect as well as the postanalytical aspect, you know, we have all of those clinics and hospitals and the courier involved, and it's just that little part at the top where the newborn screening program between the laboratory testing and follow-up program, that's the piece that -- that we, as newborn screening programs, can control. But it's incredibly important to have

communication across all of those system partners. Next slide, please.

So, with COVID-19, we've been facing a very different problem -- a very different emergency situation for the last year than anyone has ever really planned for. And there's been pandemic planning but in a lot of those situations, I know that we planned for having staff out, but we didn't plan for a lot of the situations that have been faced as far as the shortages are concerned.

And so, APHL has been in conversation with newborn screening programs regarding issues faced with the pandemic from very early on. And so, we decided to gather impact and to develop a survey so that we could get more information from newborn screening programs.

So, the results of this survey are included in the meeting materials that Mia sent out most recently. So, if you haven't had a chance to look at that yet, you can see the full results from the survey, but I'll go over briefly

a few parts of the results.

So, the survey was fielded from November 2nd to the 24th of last year. It was sent to all newborn screening programs in the states as well as District of Columbia, Puerto Rico, and Guam for a total of 53 programs. Multiple responses were allowed per state to capture the laboratory follow-up and other perspectives. So, 34 total programs responded, and that breaks down to 11 states that submitted multiple responses and 23 states who submitted a response that combined both laboratory and follow-up -- or follow-up, sorry. And so, next slide, please.

So, one of the questions asked about issues with transportation or courier, and there were a few states who noted that they didn't have any issues with transportation or courier; however, you can see from the slide that -- that there were many who experienced delays with postal service or private couriers, and one of the specific issues or some of the specific issues noted were changes with pickup and delivery

schedules or locations. Next slide.

So, other transportation or courier-related issues had to do -- and this is quotes from a couple of the surveys -- where there is a change in processes in a hospital or birthing facility that caused delays because specimens were misplaced because of that change in the process. In another situation, there were specimens that were lost because of changes in courier personnel. Next slide.

And almost every state who responded faced -- all but one who responded -- has faced some sort of staffing challenge throughout the pandemic. Many states have noted that they had staff that were redistributed to focus on COVID efforts. States have experienced staff who retired early, who changed jobs. Because of the vast demand for laboratory staff, there's been an incredible competition created for that, and so, we've experienced definitely in our lab in Texas where we've lost experienced staff because they -- they've been able to go to another company who can

pay more than we can because they're experienced and can go straight in and do COVID testing for another company.

We had staff who had problems with homeschooling and with child care since so many schools shut down and many schools are still shut down or children are still virtual -- doing virtual learning. There have been hiring freezes, which just makes the problem worse. And furloughs is a similar issue. So, next slide, please.

Some of the other staffing challenges that were noted were really low morale among staff, and we've had discussions about -- about morale and among different members of APHL. Teleworking is something that is, I would say, fairly new for almost every newborn screening program where in a lot of cases, follow-up staff especially were sent home to do their work, so they had to change processes in a very short amount of time to adapt to going, for example, from a paper-based system to paperless-type systems, even if they had -- even -- even when you

have computer systems, there's still a lot of things that were done via paper and fax that have now changed fully electronic.

Of course, there have been staff who have tested positive or family members who have tested positive, which causes the inability to work. Transportation issues when transportation has been shut down. And technology issues for those who are teleworking from home. Next slide, please.

One of the biggest impacts that we've been talking about recently and really was the impetus for this survey is some of the supply shortages that have been created, most notably there's a worldwide shortage of plastics. And so, consumables made with plastics are in really high demand, and pipette tips, in particular, have been an extreme concern for newborn screening programs. So, it's fantastic that we've implemented molecular technologies in our screening processes. But it's almost caused parts of the newborn screening programs to shut down due to that supply shortage and so, there's a lot of creativity

that's been introduced to be able to continue testing. And so, in addition to pipette tips, you know, also microtiter plates and in addition to that, even -- even the ability to get service on instruments has been limited and delayed because of the incredible demand created by the demand for COVID testing. Next slide, please.

So, even -- even when newborn screening is considered an essential service, there are different parts of the program that may be prioritized differently, and this was a quote from one of the respondents who said that even within their newborn screening program that their long term follow-up wasn't considered a priority program within newborn screening, and most of their follow-up staff had been reassigned to COVID duties, which had put an incredible strain on them to continue tracking the information that they had been working on so diligently. And they said they continued to advocate with leadership and be involved in COOP planning, but it's difficult to negotiate and change our priority level in the

middle of this pandemic response. Next slide.

So, how do we ensure that newborn screening is considered a priority and is considered essential and so, these are some of the responses that were noted. Communication with leadership through Continuity of Operations Planning, by engaging external stakeholders, and some noted just by establishing newborn screening as an essential service within the health department.

There have been concerns expressed more recently since the survey was fielded about individuals who have to work onsite, such as in the lab, and being considered essential from the stance of for the purpose of vaccine distribution. And so, again, the whole how do we -- how do we ensure that newborn screening is maintained as a priority, an essential service, and everything that should go with that? Next slide, please.

So, throughout this timeframe, as I mentioned, APHL has been talking to its members and developing resources and putting information

together. And so, this is just a list of some of the resources that have been put together. Throughout every committee and subcommittee and workgroup, there have been discussions about COVID impact, about COOP planning. APHL has reached out individually when they've heard of issues. There have been hot topics webinars that have been put together. Examples of that are things about like how do you staff during a pandemic, how do we improve the safety of our workers, managing staff during the pandemic. And their website has all of the information compiled on this as well as the Listserv that is extremely active for real-time information sharing. Next slide.

This is just a picture of the NewSTEPs website and information on COVID-19 is on the top banner, and so, it's easily located regardless of whether you have a log-in for NewSTEPs or not. Next slide.

So, I want to end by listing some of the resources that are currently available to newborn screening programs. So, the Emergency Management

Assistance Compact, again, that is what was utilized by Louisiana for assistance after Hurricane Katrina. There are state-specific health alert networks and each state has one, which may have a prescribed template that all programs must follow. Hospitals must have COOP plans. And then the Newborn Screening Saves Lives Act specifically mentions the National Contingency Plan for Newborn Screening and the most recent version, not the one that was just introduced, I haven't seen it, but the last one did have the requirement for National Newborn Screening Contingency Plan and Scott Shone will be talking about that next along with his perspective for North Carolina. And that's my last slide, thank you.

SCOTT SHONE: Great. Okay, thank you. Good afternoon, everybody, and thank you to Susan for a great overview and for telling the New Jersey story. Great job, Susan. I just wanted to -- I don't know if people can see this on my camera, but Susan showed that picture of the glass

-- the solar panel shattering. This is a piece of glass I picked up when I walked into the lab that day and took that picture. I still have it in my office as a reminder. It in no way prepared me for what we've been going through last year. But it's always a reminder of how things can go wrong.

So, for my presentation today, I'm going to wear really two hats -- committee member and speak my thoughts as to what we should do as the advisory committee with respect to the contingency planning and as a state lab director as my role here in North Carolina, and I actually was going in a chamberesque fashion wear two hats and change them on and off, but I -- as most of you know -- I'm incredibly vain about my hair, so I ditched the hats today.

And so, before I begin, I do want to acknowledge all of my colleagues in the public health labs across the country as well as all of our partners, whether they are follow-up partners throughout our Divisions of Public Health or others. This has been an unbelievable time, and I

appreciate the committee giving us time to talk about our lessons learned while we're in the middle of it as well as APHL for allowing me to speak. So, next slide, please.

Just some real quick background. Susan mentioned the Newborn Screening Saves Lives Act. While the original act from 2007 directed CDC with HRSA and state agencies to develop a National Continency Plan to be used by states, regions, or consortium of states in the event of a public health emergency. And in 2008, CDC and HRSA pulled together a workshop including federal partners, state public health programs, a variety of stakeholders to develop the initial CONPLAN Version 1, which Susan mentioned. And so, that was published in 2010. Next slide, please.

The Reauthorization Act of 2014 specifically stated that this plan needed to be updated at least every five years. This was what I alluded to this morning when Alex was talking about how often should we re-review disorders. Built into the Reauthorization Act is that we need

to assess what we're doing on a routine basis and as Susan demonstrated during her presentation, Iowa and New Jersey, after the event, evaluated what happened, what could we do better moving forward, and so, that's the same -- same concept here is it's at least every five years or really in the event of something that happens, and I think we're in the middle of that.

We're also coming up on five years, so it's kind of again serendipitous of realizing that we need to take a look at this and that this is an opportunity for the advisory committee to make comment.

So, in 2015, AMCHP was brought in to partner with CDC and HRSA as well as APHL and expert stakeholders from the newborn screening system to update this Newborn Screening Contingency Plan, and it was my pleasure to serve on that advisory committee. I believe I was selected as a result of our experiences with Hurricane Sandy, and Stan was also on their based on his experiences. But it had entire system

representation and input. You can see here public health labs in general are wonderful partners in the advocacy association, specialists, Title V, and a variety of other professional organizations, some of whom are represented on this committee.

And so, it's a very long list of contributors that I will not share. You know who you are. Many of you are on the call -- I've been scrolling through the participant list. Thank you again for all your work on CONPLAN V2. Next slide, please.

And so, essentially, our process is made up of -- we held advisory committee calls. We actually launched with a public comment survey in the winter transition from 2015 to 2016 where we encouraged people to review CONPLAN V1 and then asked a series of questions to solicit thoughts and input on how can we make adjustments and edits and changes. We actually held an in-person working meeting -- remember what those in-person meetings used to be like -- and so, we broke up, and it was really divide and conquer. We had

subcommittees that took apart different pieces of the CONPLAN and began to develop resources for that, and that was really comprised of what you see on the graphic on the right, which was four main -- there were four main foci of -- of update. We looked our strategic objectives for the plan. They were updated. So, they were tweaked. We added -- we also reordered them to be more functional in terms of chronology of the newborn screening process but also a level of just thinking as you think through a newborn screening system.

Point of care was woefully left out, largely because how early the CONPLAN 1 was drafted. So, we assured that EDHI was represented strongly, but also CCHD was added. We certainly need to make sure that our point of care newborn screening tests are included in all of this. This is not just about assuring dry blood clot testing, but all these critical first tests that these babies get when they're born.

We updated the responsibility matrix, and

I will just say here, reflecting on the CONPLAN for this -- for this talk, it occurs to me that we probably as a committee added people to the matrix and added responsibilities without really them fully grasping that they now had these responsibilities. And so, lessons learned moving forward is not only do we have to incorporate all these people but really make it clear as what are their roles when this happens. And so, the responsibility matrix was a great idea, and I think really well drafted. But again, closing the loop on that is making sure that everybody in the matrix understands that.

We updated our appendices and what I thought was great was we developed new resources, which I will elaborate on in future -- in three or four slides. We submitted -- the advisory committee submitted all of these revision recommendations to our federal partners. It underwent a thorough CDC and HRSA review and actually was published in August of 2017. Next slide, please.

I'm going to go into the strategic objectives really quickly and talk about they encompass the whole process of newborn screening from communication, education, assuring that there's a framework for blood spot, hearing, and CCHD, specimen collection, and transport as needed to the laboratory, testing, results reporting, diagnostic follow-up, treatment management, and then obviously anything else we think of that needs to happen.

And Susan sort of foreshadowed a little bit of my thought here which is that of all of these bullets, only two really rely solely on the newborn screening programs, right? The laboratory and follow-up. That is specimens are processed and tested and that the results are reported and followed up accordingly.

The rest of this, while the programs have a role in the other bullets, the rest of this involves the entire system. Susan's slide -- and I'm going to use that graphic a little later -- but just to continue with her comment, the most

effective continuity plan and preparedness is going to rely on way more than our newborn screening programs. Next slide, please.

This is a great infographic. I'm hoping it's visible. If it's not, I will reflect on Sue Barry, who put up a wonderful metabolic pathway several years ago and said, "I put this up to impress you, not for you to read it." I actually hope you can read this. And so, you know, across the top is our communication strategy and assuring that communication happens. Joyal Meyer and Michele Caggana are speaking after me. Michele is going to talk, I think, about some communication challenges during the pandemic and how in the middle there, a plan for communications to all stakeholders during emergency event is established. Programs can't out that hospitals close their L&D through a newspaper or through a news alert, you know, in response. There's a cascade of communication throughout the system that has to happen and, you know, during Sandy, we identified that our partner, who was leading

communication, was doing a great job of communicating but not to the right people. And so, you know, it's critical in terms of this understanding.

Education is often like sort of thrown out. That brochure is just thrown out. But I think we need to think about how do we tweak education when there are so many other competing priorities in an emergency to make sure that we're still communicating effectively and accurately.

And there's a whole host of -- down the columns here -- these enabling functions that go with of the strategic objectives. And I'll just reiterate again that the yellow and blue in the middle are really where the state -- where the state lags -- rather the programs are focused -- processing samples, reporting results. I think we, as an advisory committee, need to look across this whole infographic and think about where do we -- on the committee as well as organizational reps -- fall in assuring that all of this continues during an emergency, and are we prepared for that

process. Next slide, please.

We -- the advisory committee for the rewrite or the revisions for the CONPLAN 2 were focused on making sure that the plan was usable -- that it wasn't just a document that said you need to have a continuity plan and these are all the things that should be in it and that's it, because largely, that just sits as a binder on somebody's bookshelf gathering dust. We deliberately talked about what would help ease use in the event of -- let me back up -- ease use to prepare and set up for preparedness but also in the event of an emergency.

And so, we tried to really facilitate uptake and that was intended for people who were developing COOPS or who were revisiting their existing ones and looking further. So, EMAC -- Susan mentioned EMAC. So, EMAC was a big part of our -- of the CONPLAN V2 and how to employ and use it and prepare for its use. We also had a variety of MOUs that other states had used to share. We addressed the common themes that people often

thought of. This state has a different disorder panel than me. How are we going to pay for things? We deliberately addressed these hot topics to help people think through how to get through that, right, because when push comes to shove and you need to make sure babies are screened for these critical time-sensitive and time-critical disorders, we need a path to make sure that that happens, right? I will say that not once during the deliberations on the advisory committee did we ever think about a global pandemic that impacted every single person at the same time. And hindsight is 20/20, but the good news is we have all lived this, and my last slide, I'll talk about how I think we need to revisit CONPLAN 2 for 3 and begin to think about pandemic planning in the scope of newborn screening and public health. Next slide, please.

Kate Taft from AMCHP presented to the advisory committee in 2017 and in the 2017 -- the 2013 to 2017 advisory committee report to Congress, the existence of this plan was

mentioned. It passed HRSA and CDC clearance. The plan was always what I call to get the band back together. We were going to bring back the advisory committee for the CONPLAN to come up with a dissemination plan to advertise it's published and also to develop review and revisions. And we really didn't do that. APHL did a great job of announcing it's existence, CDC pushed this out. The fact that it existed again, I think, was well known. There were presentations at the Newborn Screening Symposia, as I said, at the advisory committee itself. But that last bullet about the intention to work with programs to develop, implement, and maintain, I think is a failure of the system. We just did not do this. Me neither, right? In my role in New Jersey or my role here in North Carolina, we have not yet really taken this and put it to use, and that is the challenge that we need to have -- that we need to take on as a system to improve.

So, if we go to the next slide, I'm going to put on my lab director hat, and as I said, I

don't have a hat, but I'll hold up my North Carolina Flag pin. And so, as the North Carolina State Lab Director, I went to our Newborn Screening Program manager, Kimberly Blake, and I said Kimberly, okay, what is your perspective of COVID-19 and how it impacted us here. I think we had a lot of successes. Carla mentioned earlier we need to celebrate our successes as well as acknowledge our challenges. So, I think we -- we did a fairly good job responding. There obviously were challenges where we split up the staff in two early on, you know, pre-masking, pre-plexiglass, pre-reorganizing the lab. We were just, all right, half of you are here on one day, half of you are here three days later so that we don't have overlap and so, there were those challenges. But I think it helped us avoid significant exposure, people being out. Obviously, everybody had our challenges.

But some of the themes have happened in North Carolina, which Susan alluded to in the survey, reallocation of resources to other areas.

I will say that inevitably, every time when I would ask our team what are we doing, it was always, how is the COVID team, how is newborn screening? Obviously biased based on my history and my current role, but -- but that was important. We weren't diverting pipette tips from newborn screening to the COVID team, but we understood that vendors were reallocating based on COVID, not on newborn screening, and so that was -- that was something that we had to address. So, supply chain was a big issue.

Our courier was a problem. We had it impacted by staff exposures and obviously volume delays, and we all fell victim to USPS, especially in October, November, December, and I'd stay it's still ongoing. I have staff who just got Christmas cards that were mailed in December. So, there's still challenges, I think, in our courier system depending on what courier you use for what aspect of your -- of your program.

And finally, project management -- and we had resource reallocation. Our IT team was having

to configure multiple COVID tests, was having to respond to adding new instrumentation, our facilities team was making sure that we were adding all of these new instruments and all of these new pathways for COVID, and we had to make sure that we were still in the process of adding disorders to our newborn screening team, right? So, our program is undergoing a massive expansion to bring us congruent with the RUSP this year and so, our resources internally to the state lab here were redirected mostly on the human scale.

I also asked Kimberly, okay, what else, and I put her quote here. She said, "Business decision makers were otherwise occupied," which to me meant Scott, your attention was drifted elsewhere and when we needed you to make a decision as lab director, you, unfortunately, weren't able to. And rightfully so, I mean, all of us are spending 12 to 16 hours a day with 99 percent of that was on COVID for a long period of time, but it -- I think it's a real eye-opener to leadership on how much we have to pay attention to

this, and I've been a lot more deliberately hopefully now, that we can continue to move things forward.

So, it's important to remember that success or everything thinks it's this straight line, but even without a pandemic, it takes a long circuitous route to get there and the pandemic just adds a whole bunch of more squigglies. Next slide, please.

And so, I want to end my last two slides on how I think we can move forward, right? So, I look at this picture a lot and think, oh, I feel like the truck. But I think this is how programs feel -- newborn screening programs, metabolic genetic programs, pediatric endocrinology programs, you know, we all have this going on. Pre-pandemic, maybe half as many people on the truck. But now, it is -- there's just an overwhelming amount of things to tackle, right? But -- next click.

We operated like freight trains, especially newborn screening programs. We just

go. We barrel -- we find solutions. We address timeliness. We add disorders. We address unsatisfactory samples. Follow-up finds the parent that is missing. You know, we trug along and barrel through anything in our way because we are freight trains. The problem is there is an enormous inertia to adding cars onto this train. And that's the next slide.

We need train cranes. I practiced saying that all last night, and I did it right. We need cranes to help put more cars on the program tracks. So, next slide.

So, what do I think we need to do moving forward? First of all, assuring newborn screening is part of our public health lessons learned from the COVID response. There is a tremendous amount of energy, effort, money going into being prepared for the next infectious disease, viral pandemic, et cetera. We have to look at our public health system, which has been exploited to be so well worn, inequitable, inefficient, and we have to make sure that newborn screening is part of the

discussion of how are we going to fix this with the resources that are coming in. We need to identify cranes. Programs are not going to be able to do this. This is not another pile on top of that truck. We have to work together and deliberately work with programs for system improvement to make sure that we can implement plans and therefore be prepared when this happens again. And planning is not preparedness. We need to exercise, we need to learn, in between these events, we need to keep this process of continuous quality improvement ongoing. And it has to be a system approach. It has to be. Again, programs can't do this alone -- any programs. I'm obviously very protective of our newborn screening program, but every program has to work together to make this happen.

And so, my challenges as an advisory committee member, Dr. Powell, is that we have to figure out to communicate with the Secretary, to whom we advise, to make sure that newborn screening remains part of HHS's efforts to improve

the Public Health Response System moving forward. Whether that's through mechanisms that aren't currently focused on newborn screening -- obviously, there's a lot of money coming in through the ELC grants -- or whether we revisit PHEP, the Public Health Emergency Preparedness Process. I had great success when I was in New Jersey to get a modicum of funds when that was a key focus of PHEP. But we need to explore and think as big and as diverse as a pandemic on how we're going to respond. And that's my last slide.

I really appreciate it and I am so grateful for the talks coming up from our partners at -- in Nebraska and New York. Thank you.

JOYAL MEYER: Thank you so much, Scott. Thank you everyone for having me here today. My name is Joyal Meyer. Can you guys hear me okay? Susan, I can see you. Yep, okay. Awesome.

My name is Joyal Meyer, and I'm the Newborn Screening Program Director for the North Dakota Newborn Screening Program. So, today I'm going to be providing an overview on our program

here in North Dakota. Next slide, please.

So, I just wanted to provide an overview from a North Dakota perspective. And so, the total population that we have in North Dakota in 2019 was 762,000, and so, the population of people per square mile is 9.7. Only Montana, Wyoming, and Alaska have a smaller population per square mile. So, there are 39.1 million acres and nearly 90 percent of North Dakota's lands are farms and ranches. North Dakota is the top producer in the nation of spring durum wheat, dried peas, beans, flax seed, canola, and honey. And so, North Dakota is also the number one producer of honey, and so a fun fact that last spring, honey bee workers needed actually -- they needed to have quarantine letters -- exemption letters at the beginning of COVID to be able to transport the bees back to North Dakota from California to their winter home. So, next slide.

So, as you can see from these pictures, and if you can fast forward two times, please. There you go, thank you. As you can see from

these pictures, the weather in North Dakota is very unpredictable. And so, the picture on the left there is actually one of our malls here in Bismarck, which is the capital of North Dakota mall parking lot, where you can see the tops of some of those cars. And the picture on the right was taken a few years ago off of Interstate 94. And so, we have very unpredictable weather. The high for today is -9 degrees with a wind chill of --makes it feel like 33 below. And so, we have had snowfall in June, and so the cold temperatures, unexpected blizzards, and poor road conditions can make for some challenging times for our courier service in North Dakota. Next slide, please.

So, in North Dakota, we have 28 independent local public health agencies, and some of these are combined, as you can see on the map. So, 75 percent of the local public health systems and units serve as a single city or combined city and county jurisdiction, while 25 percent serve the multicounty jurisdictions. And so, the

majority of the multicounty jurisdictions shown in the light brown reside in the western portion of our state. The public health units are required to meet standards and follow the state laws and regulations, but they do exercise their own powers and have administrative authorities to make decisions that meet their local needs. Next slide, please.

So, there are approximately 12,000 babies born each year in North Dakota. We have 12 birthing facilities and out of the 12 birthing facilities, 4 of the 12 approximately have 60 percent of our total births in North Dakota. Two of the main facilities are located in central North Dakota and 2 are located in the eastern portion of the state. Next slide.

So, this graph shows the occurrent births that we had in North Dakota from 2011 until 2019, and so, this includes all babies born in North Dakota, so not just the North Dakota residents. And, as you can see, from 2011 to 2016, the birth rate increased every year with a peak in 2016

being 13,035 births. So, this was an exponential growth, which was a result of the oil boom in western North Dakota, and so individuals and their families relocated to North Dakota for jobs in the oil industry. Next slide.

In North Dakota, we also have 5 federally recognized tribes and 1 Indian community, which are shown in yellow on this map. There are 31,329 American Indians that live in North Dakota and they make up 4.9 percent of our total population. Sixty percent of those 4.9 percent live on reservations, and over 40 percent of these American Indians are under 20 years of age.

And so, with the government systems, they have no authority -- we have no authority to track down children and families on the reservations because they function independently from the federal and state government. So, each tribe has its own constitution and set of laws to govern and conduct with -- that they govern and conduct with the jurisdiction. So, at times, this has posed some challenges in locating babies that we've had

with abnormal screens that reside on reservations in North Dakota. Next slide.

So, as the population in North Dakota has grown, the diversity in North Dakota has also increased. And so, this graph shows the number of births to foreign-born women who are North Dakota residents from 2009 to 2018. And so, 11 percent of the total births in 2018 in North Dakota were to foreign-born women, and over the past 10 years, women from 176 different countries have given birth to babies in North Dakota. Next slide.

So, this slide illustrates that as the number of births have increased in North Dakota, so have the number of confirmed traits and disorders. In 2019, there was a significant rise in confirmed traits and disorders. And so, this included an increase of hemoglobin and cystic fibrosis traits and babies diagnosed with cystic fibrosis. Next slide.

The University of Iowa State Hygienic Library began screening for newborn screening in North Dakota in 1992 and also in 2007 nurses at

the University of Iowa hospitals and clinics began doing short-term follow-up services for North Dakota. North Dakota and Iowa currently have a memorandum of understanding for the laboratory screening and for the follow-up services that they provide. North Dakota began outsourcing newborn screening in the '90s because of the increasing fees of equipment and laboratory costs. It was also more feasible to partner with Iowa when we began screening for metabolic disorders since they had the expertise and infrastructure to do so.

In addition to North Dakota and Iowa, the State Hygienic Lab also provides screening and follow-up services to South Dakota and Alaska, and Iowa processes the billing for North Dakota newborn screens and invoices North Dakota facilities who in turn bill the patient's insurance.

And just as an FYI, the current newborn screening fee in North Dakota is $96.00, and that includes lab processing, short-term follow-up services, which is provided by Iowa, the courier

service, Iowa physicians also provide medical consultation as a backup for our North Dakota specialists on the evenings, weekends, and holidays. Next slide, please.

So, we have a courier system, and our courier is called Meadowlark Logistics, and they have transported specimens for us and they have transported specimens from North Dakota to Iowa 7 days per week, 365 days per year since July 1st of 2019. So, prior to that, we only had transportation 5 days per week for the birthing facilities and some of the larger facilities we did have transportation available on Saturdays. And so, there was no transportation available on Sundays, even though the State Hygienic Lab was open 7 days a week.

During the weekdays, specimens are transported via ground transportation and then flow into Fargo, North Dakota where they are transported from Fargo to Minneapolis, and they are combined then with the South Dakota specimens and driven to Iowa. So, it's quite the process

for our specimens, but it works out great.

During the weekends, the specimens are transported strictly by ground transportation, and in the case of an emergency, our courier system can transport specimens on any charter plane if they need to. And so, they have many shared customers and close relationships in our surrounding states. Next slide.

This graph is courtesy of the NewSTEPs data repository and it includes timeliness data from 2015 through 2020. And so, as you can see, since 2016, more than 95 percent of our specimens collected had time-critical results that have been reported out by 5 days of life. And so, this high percentage is really a result of our continuous education to our 12 birthing facilities, the outstanding courier service that we have, and the work of our partners in the Iowa lab and short-term follow-up programs. So, we certainly couldn't do it without all of our partners. Next slide.

So, in the Newborn Screening Century Code

for North Dakota, it's mandated that the program provide education to licensed clinicians, hospital staff, public health nurses, and citizens of the state. And so, in the fall of 2015, we actually began having in-person trainings with all of our 12 birthing facilities and so, these visits have not only benefited the hospital, but also us as a program, and we have found that, you know, we found our newborn screening champions during these visits and have really developed relationships and rapport with the nurses and the lab staff there.

We have created education modules that provide an overview of the newborn screening programs and disorders that are included in our panel. We've hosted hot topic lunch and learn sessions for our partners. We've had a few -- two newborn screening conferences in North Dakota and had great support and attendance at both of those events that we've had.

We also have an advisory committee that meets on a quarterly basis, and we are fortunate to have representation from all of our 12 birthing

facilities on this committee as well as our travel partners, parents, and family support groups. Next slide, please.

So, after we [inaudible - audio cut out] the hospitals, the percent of our poor-quality specimens decreased significantly. So, this graph is also courtesy of the NewSTEPs data repository, and it includes the number of unsatisfactory specimens collected from 2014 through 2020.

And so, North Dakota has been fairly stable with poor-quality specimen, below 1 percent, until the beginning of COVID last spring, where we experienced an increase of poor-quality samples similar to other state newborn screening programs.

The poor-quality rate has since declined and we have continued to provide education to hospitals and clinic staff on a routine or as-needed basis virtually. Next slide.

North Dakota is a member of the Heartland Regional Genetics Collaborative as well as other states that are listed on this slide. And so, the

goal of this network is to increase genetic services, particularly for medically underserved populations, and we do this by helping to increase accessibility to care by enhancing telehealth and teleogenetic services that are offered to parents or patients and their families while maintaining that high quality of care.

In North Dakota, our only metabolic geneticist is located in Fargo, which is on the eastern part of the state. So, this has posed some challenges for families, especially those that live on the western part of the state. And so, telehealth has really been a great opportunity for us to connect with those families, especially during the -- through COVID and during our winter months. Next slide.

So, though North Dakota -- the North Dakota Newborn Screening Program does not have a formal contingency plan, at this time, we are planning to partner with the other states who also use Iowa as their screening laboratory. And since Iowa processes North Dakota newborn screening

specimens, the North Dakota State Lab also has had a minimum -- has had minimal involvement in our program. The state lab and the newborn screening program utilize the same courier service, and so we -- as we move forward in developing our COOP plan, we plan to include our state lab in the planning to help streamline our processes.

So, I wanted to mention a few things about the state lab that we have in North Dakota. They are a member of the Northern Plain Consortia, which includes North Dakota, South Dakota, Idaho, Montana, and Wyoming. And so, prior to COVID, there were 18 employees that were at the state lab, and that included laboratory and support staff. And so, after COVID, the number of laboratory and professional support staff increased to 140 full-time and part-time employees. So, it went from 18 to 140 and at the peak of testing, they were at 140 full-time employees. So, they had to increase their support significantly.

Prior to COVID, the state lab in North

Dakota was -- the operating hours were 8 to 5 with no weekends and no holidays, and then after COVID started, the operating hours are 24 hours a day currently and 7 days a week, and this includes weekends and holidays.

The courier transportation prior to COVID was 5 days a week and now they currently have 7 days a week. Similar to other state labs, their lab has also noted limited supplies and testing for them.

So, for our newborn screening program, we do have 2 FTEs at the state level, so it's myself and another nurse coordinator, and we have been a part of the COVID response since the -- on behalf of the Department of Health since March of last year. So, just this week on Monday, I was actually relieved of my COVID response duties, so I am happy to be moving on and back to newborn screening work. So, my coworker, Amy, is still assisting with the COVID response, and we're hoping in the next few weeks that we will both be solely working on newborn screening response --

newborn screening again. So, if there's another surge in cases, we will need to assist with those -- that response.

As far as what is on the horizon, if you go to the next slide please, the North Dakota Newborn Screening Program began doing long term follow-up in North Dakota back in 2019 and so, we've been working for the past couple of months to develop a system and a data base that we can track and follow newborns after they have a confirmed disorder. So, we were recipients of the APHL Continuous Quality Improvement Grant and so, we've been really working closely with our health information staff and IT staff to work through some datapoints that we want to capture and see where it's best to store that data at for easily accessible information for our providers.

The North Dakota Newborn Screening Program welcomes future discussions on any contingency planning, and we would be happy to be involved in future meetings related to this topic.

And thank you for allowing me to share

our perspective on newborn screening in North Dakota. You can go to the next slide.

And that has my contact information. If you have any questions, I'm happy to answer.

MICHELE CAGGANA: Hi everyone. Can you hear me? Okay. My name is Michele Caggana. On behalf of myself and [indiscernible 1:55] members, I want to thank you for your interest in this topic. It has taken a lot of our time over the last year and allowing the opportunity to share our experience. You will definitely hear some common themes throughout my talk this afternoon. Next slide, please.

So, interestingly, people always think of spring as a time for rebirth. But in newborn screening, we're interested in all the births. And in early March 2020, we became aware of several changes that were happening in New York City, which I'm going to discuss in this presentation. These things included the possibility of early discharges from hospital, changing availability of specialists and

providers, as some were deployed to emergency departments or told to close their offices and do telehealth. We also received reports from downstate of parental hesitancy in bringing their babies on public transport or to the hospital and even to their pediatric doctor or clinic. We heard about many provider clinics and offices closing as well.

So, in hearing these issues, we realized that we had to maintain our operations and potentially with greatly reduced staff, and we were also concerned about our own staff, their well-being, and their families also. Next slide.

So, beginning on early March, Beth Vogel, Joe Orsini, and myself sprang into action because we knew we had to think outside the box and make some changes really quickly. So, one of the benefits of this, which kind of is a theme throughout this talk, is that we were already working on some efficiencies in our program and COVID actually accelerated the implementation of these measures. And so, we did get a benefit out

of it. We engaged our staff for the perspectives because we realized that we would need to ramp up our COOP planning in the event that many of our staff became ill, and we had to maintain operations because we were a mandated public health program. And so, we had to screen. We couldn't afford to not screen specimens that were coming in. Next slide.

So, we decided to look at the entire newborn screening system from our perspective, and I'm going to review what we accomplished and the things that we did in each of these areas of the laboratory newborn screening in general. Next.

So, first and most importantly, we had to consider what our outside challenges were going to be. As early as March 2nd, we began getting phones calls from our New York City Hospital Newborn Coordinators that parents were refusing to come in to get repeat specimens collected and that their outpatient clinics were closing. We also were told the hospital administration was telling the labor and delivery departments that the babies

were not -- the families were not going to be allowed to come back to the nursery for any specimen collections and that some of the pickup locations for newborn screening specimens were changing. Some hospitals even reported that the couriers weren't allowed to enter the hospital, let alone come up to the floor to pick up specimens. So, we had to work with UPS to change the pickup locations so the drivers knew where they could go in the hospitals in order to pick up the specimens.

Of special concern, the hospitals were telling the nurseries that they were going to release babies and their mothers as soon as possible within 12 to 24 hours after delivery and this posed somewhat of a problem for us and spawned a lot of data analysis quickly on our end, because our references ranges were all set up for babies who were greater than 24 hours of age when the specimen was collected. As I'll show later, luckily the neonatologists were successful in pushing back on this.

We also learned that some of our specialty care center staff were being redeployed to emergency departments and ICUs and they were also ultimately told to embrace telemedicine. So, they were told their hospital offices were closing.

Our newborn screening coordinators told us that they could no longer process paperwork or accept any of our phone calls about collection of repeat specimens, and we've been working on doing E-mail communication with them, so this became our main mode of communication.

And as you probably know, our governor first called to reduce essential workforce by 50 percent in mid-March and just two weeks later, we entered a full New York state pause, and with that, New York basically closed down. Next slide.

So, as far as accessioning is concerned, we put a protocol in place to separate specimens from our COVID-positive mothers, and we put them into their own batches on our tally. And we did this in case we would need to pull them later on

for any kind of study, and also to prevent our data entry staff from having to handle them.

We sent an E-mail blast of almost 10,000 out to our health care provider E-mail list informing them of the various changes that were going on. We notified them that the provider offices might need to collect repeats -- this is something they weren't necessarily accustomed to doing -- and that they might have to actually manage referrals by themselves. So, we included a distribution of information and education about specimen collection and information on how to order forms. And we also put together fact sheets for the various newborn screening conditions that went out so that the providers would know what to do based on the fact sheets if they had to actually work on a referral for a metabolic condition on their own.

It was good to see that we saw a rise in requests for newborn screening collection forms. So, at least we know that the providers actually read the E-mail. Most of our providers only keep

a few forms on hand because they're not used to collecting repeat specimens, and so they heeded the warning and actually ordered collection forms.

We also initiated Saturday testing in order to increase the time that might be needed for us to find babies with critical screening results knowing that parents were hesitant to answer phones and bring babies in, and it also helped us to manage our daily workflow, you know, anticipating that some of our staff would become ill and we would have a decreased number of people to actually do the lab work piece.

The newborn screening program also helped with COVID sero-prevalent studies, and I'll talk a little bit more about that. These were offered to the public in New York and essential workers and others and we put our high throughput skills to work to help accession specimens for the sero-prevalent studies. Next slide.

Our data entry staff were moved to another building on Friday, the 13th of March, of all days, and we did this in order to decrease

density and free up space for COVID testing and accessioning. So, newborn screening kind of condensed down to allow expansion of testing for the virus if it was necessary for the state. And these decisions were all made very quickly.

We also implemented an all-hands-on-deck data entry mode. So, what we ended up doing was scanning packs of the newborn screening demographic forms. We created an electronic checkout sheet so that people could work remotely, if necessary, and check out a pack and do the data entry, so we would know who was working on what piece. It also enabled staff to begin doing data entry even if they were off site as soon as accessioning was complete each day. We had two students who worked nights and weekends because they were taking remote classes, and they helped to keep up caught up very well.

Just a few days later after we moved the staff out of here to another building, two weeks later, we set up to offer remote staff -- remote work to administrative and grant-funded staff,

again to decrease the density onsite. Next slide.

Follow-up was very busy as well, as I mentioned. They were also moved on March 13th to decrease density, and our IT staff person, Chris, quickly set up -- and our IT staff at the center -- set up remote access for our staff. Most of follow-up did go remote by March 30th, and all but one staff working remotely. And this was a big accomplishment because, as mentioned, it's something that newborn screening just wasn't accustomed to -- being able to do work remotely.

We also spent a lot of time with the educational piece that I discussed, and we also updated the language on our reports to make them more descriptive. So, Beth Vogel pulled a lot of data and we reviewed the information and ended up creating risk-based language for our borderline results. So, for example, instead of the banner reading to collect a repeat as soon as possible that it was a positive newborn screen, now it reads something like 1 in 10 infants with this result for PKU were diagnosed with classical PKU

and then it says, please collect a repeat as soon as practical. So, in this way, the provider was given information to determine how to promptly get the family so they had that information to see like how urgent they would get that family in to collect the repeat specimen.

In addition, we added TPN language for multiple analytes when they were out of range, and we decided to refer babies based on the regular referral levels when their specimens were suboptimal.

Prior to COVID, we only referred out infants who had a suboptimal specimen collected when their results were at the emergency or critical level that we typically use for weekend callouts. So, this was one of those pipeline changes that I spoke about earlier that we were able to do because COVID was with us.

We also sent E-mails out to all of our specialty care center directors and their staff, and we asked them to update their contact information and let us know if there were any

changes to their staff as well. And we had a call with our CF Specialty Care Center directors to review the CF Foundation's COVID guidelines and told the centers to review any issues related to CF disease and the issues with COVID.

Because the New York State sero-prevalent study was ongoing and they needed quick access to cards to be able to collect blood on finger sticks, our newborn screening forms were used in the early stages of the sero-prevalent study for the public. So, we fielded some very entertaining phone calls from the public looking for their COVID results because, of course, they had our phone number because people at the collection sites gave them the pink copy of our collection forms, which we typically give to parents of newborns. And so, our phone number was on there, and they ended up calling us, and we had some good laughs about some of those voice messages. Next slide, please.

In the laboratory, as I mentioned, we tested on Saturday and we began the remote data

entry, and all of our lab staff helped with that as well. We were able to analyze all of our data for lysosomal storage diseases, for tyrosinemia, and adrenoleukodystrophy, and this continued on an as-needed basis and we no longer do the Saturday testing. At the time, the staff were very willing to come in and help with this.

We also set up -- and this was a huge lift -- we set up our instrumentation for remote data analysis, and this was in the event that we had very limited testing staff on hand. And so, we thought that if staff were well enough, if they were home because they were ill or in quarantine but they were well enough, they could work from home to do data analysis as if they were sitting right at the instrument, and we still have staff doing this for some of our sections.

We also reassigned some of our staff to maximize the timeliness for referrals and a separate area was set up for pulling repeat specimens to control density in the accessioning area.

We cross-trained some of our own staff, and this is an ongoing effort that we're working on. But we also pulled in some nonessential staff from the environmental mass-spec lab, and that was in the event that we were short-staffed in the newborn screening program so that we had additional people who were not within our program that would be able to help out. These folks were also trained on mail opening and punching as well, and we also made our GALT testing a semiquantitative test and streamlined the assay so that more people could be trained on it. Because GALT is time critical, we wanted to make sure we would be able to get that done, and previously calling out GALT results was done -- it was more of a subjective call rather than a semiquantitative call. So, this was also another thing that was in the pipeline that we were able to put into place due to COVID.

We also kept in contact with our vendors on supplies and equipment. We had some issues with service people being able to come in, and we

had some equipment that was ordered but delayed due to worldwide shortages on parts and staffing because staff were required on their end to assemble the instruments. So, we did have some delays there.

And then we began to hear of early -- pretty early on of intermittent shortages in lab supplies, and that was talked about -- tips and gloves as well -- and this occurred as, you know, as COVID testing ramped up, supplies became more sparse for the newborn screening program. Next slide.

So, basically, the take-home message was that we realized we really needed everybody to chip in in order to manage our workload and still remain functional. So, we -- we typically looked at where we needed help and solicited help from staff. Some of our newborn screening staff actually worked on the COVID accessioning for the viral testing after their shifts and on weekends in newborn, you know, the weekend shifts for COVID after newborn screening, and this gave us -- some

of our staff some scheduling flexibility as well. And, as I mentioned earlier, we helped accession the sero-prevalent samples and on one morning, we accessioned 3,700 specimens prior to 8:30 a.m. when the newborn screening mail arrived. And that meant opening packages, looking at quality, and also getting them punched. So, that was, you know, people were really proud of that.

Because newborn screening is accustomed to high throughput work, our staff were very happy to share their expertise, and they felt like they were helping.

Chris, our IT person, has been working remotely and has implemented all the reporting changes that we needed and any other necessary changes and assisted with getting people onboarded for remote work.

In general, we report to work with a temperature check. Now, it can be done at home -- previously, it was done onsite for everyone -- and we went from having masks requested to mandatory. And, of course, as you all know, any in-person

meetings or events remain on hold. Any meetings that are held in person are held in very small numbers in very big rooms, and WebEx is our new best friend, and we're able to get most of our staff web cams so that we can actually see them when we're having some meetings. Next slide.

So, I think Scott mentioned, we find things when we actually look at data. So, here's an example illustrating how we learn that a labor and delivery department actually closed. So, the dots on this graph are bursts from -- and time is on the bottom, so the dates are on the bottom -- and you can clearly tell that we abruptly stopped receiving some specimens from one hospital, which are the pink dots, but then other hospitals, we saw a big uptick in specimens or the purple dots. So, we thought something was up and our first fear was that something -- some samples were lost somewhere along the way. But the translation here is that apparently unbeknownst to us, arrangements were made by the hospital with the pink dots to send their babies for delivery to the hospital

with the purple dots and we never were told this occurred but we realized that when we collected our own data and called the hospital and said hey, we noticed this, what's going on, and our suspicions were confirmed that basically one hospital had shut down.

We were told that down state hospitals -- and it was on the news and all over the place -- but some of our down state hospitals totally reorganized their workflow in order to handle COVID patients and create COVID-specific floors. And so, this is sort of an example of that in real time. Next slide.

And as I mentioned, we were hearing that hospitals were being told that the labor and delivery departments were being told that they were going to discharge their babies early. And so, we plotted this data -- Denise Kay collects this data -- and this hasn't really changed here, but we didn't -- we didn't see a rise in very early collections -- the 0 to say 12-24 hour collections, but you definitely can appreciate in

green here the uptick in samples that were collected between 24 and 26 hours after birth. And the good news is this actually sustains over time. So, this trend remains as you see on this graph here and, in fact, when you're looking at newborn specimens coming into the program, you can see that many of them are collected at 24 hours and 1 minutes after the baby was born. So, there still is this impetus to move people through labor and delivery. Next slide.

We also did not see a rise in the time to collect repeat specimens, as plotted on this graph, because this is one of our main concerns. And so, kudos to the providers in New York who found a way to make sure that they found these kids and got their repeats collected in a timely fashion. Next slide.

That famous jellybean diagram here to reiterate the previous discussions here. What I hope that I've tried to show you today is that it makes -- this presentation makes it very obvious that COOP planning takes on many players, just as

the newborn screening system has many players. In creating all of these changes, we had to rely on providers, clinics, hospitals, labor and delivery, couriers, care coordinators, specialists, our own staff, parents, and others to ensure that no babies fell through the cracks. While we can plan for lens outage or changes in test reagent availability or fast-moving ice or snow storm or flood or other localized weather challenge, an event that takes out half the state or an entire state is very difficult to plan for. Because necessity is the mother of invention, we had to quickly pivot in our program and ramp up to remote work, cross-train staff, and sustain operations.

So, now we are dealing with, as was mentioned, the supply chain shortages that you heard about and I'll also put gloves on that list of tips and plastics as well.

These changes have led us to reformat some of our testing, specifically with tip usage, to ensure that we don't run out of reagents. So, as the survey showed, newborn screening programs

are dealing with increases in staff departures -- these could be early retirements or changes in jobs coupled with hiring freezes. There's also budget shortfalls and other issues that we have to reconcile while maintaining our operations.

And so, while the focus has changed, COVID-related COOP is still evolving. Next slide.

But at the end of the day, teamwork was key in our efforts to keep screening babies in New York. So, I want to give a shoutout to our staff. As COVID began and continues, we were and continue to be exhausted, but we also were impressed with providers across the state, our own staff, and our newborn screening coordinators. We were invigorated too because, as I mentioned, COVID gave us the ability to make needed changes in our program and we have settled into a new normal.

So, our newborn screening superhero, shown here -- we have versions you can get in color if you want -- but anyway, it hangs on the walls of our floor here at the program, and it's compliments of Shane Moore, who is the husband of

our follow-up supervisor, Sarah Bradley. So, thank you for your attention.

CYNTHIA POWELL: Thank you. Thank you all for your excellent presentations. Thank you for all the work you've done throughout this pandemic. And, unfortunately, we won't have time for discussion, but we will take up this topic at future committee meetings.

Before we break up into the workgroups, I just wanted to go over the discussion questions for the three workgroups.

For Education and Training, what type of information and educational resources would be most helpful when a conditions is added to the RUSP? What range of issues related to education should the advisory committee consider when a condition is added to the RUSP?

For Follow-up and Treatment, your questions are: What type of long term follow-up information should be considered when a condition is added to the RUSP? What type of information should be considered in a systematic review of

conditions on the RUSP? Should the cost of treatment be a factor in both the nomination process and the review of conditions on the RUSP?

And finally, for Laboratory Standards and Procedures, what information would be most helpful from newborn screening labs related to the review of conditions on the RUSP? How can we prepared newborn screening labs to collect and report this data? Should there be more in-depth information regarding cost to labs for adding a new condition to the panel, or is there already enough information provided?

And finally, for all workgroups, in your workgroup meetings, please also discuss if there are any other considerations for enhancing either the nomination process or review of conditions on the RUSP.

And I'll now turn it back over to Mia, who will briefly review instructions for logging into the workgroup meetings.

MIA MORRISON: Thank you, Dr. Powell. And just before I begin the instructions, LRG,

this slide seems to be a little bit out of focus. So, folks may not be able to read the URL.

So, you will need to log out of this webinar to log into your workgroup Zoom meeting. Workgroup members, you should have received a link to the workgroup meeting page via E-mail. This link -- through this link, you can access each of the three workgroup meeting pages.

Nonmembers, if you are interested in attending a workgroup meeting, you can type this website into your browser and also LRG should be putting that link into your chat box, and you can click directly on that link to access the workgroup meeting page where you can select the workgroup that you'd like to attend.

And just as a friendly reminder, please note that if you are a nonmember attending a workgroup meeting, you may only speak if called upon by the workgroup chair or co-chair. Thank you.

CYNTHIA POWELL: Thank you, Mia. That concludes our meeting for today. We'll reconvene

tomorrow morning at 10 a.m. eastern time. Thank you.

[Whereupon the meeting was adjourned.]

[Off the record at 2:35 p.m.]