Health Resources and Services Administration

Advisory Committee on Heritable Disorders in Newborns and Children

Meeting by Webinar

9:30 a.m. to 11:27 a.m.

Tuesday, September 24, 2019

Reported by: Ashleigh Simmons

PRESENT

ADVISORY COMMITTEE MEMBERS

Cynthia M. Powell, M.D. (Chairperson)
Professor of Pediatrics and Genetics
Director, Medical Genetics Residency Program
Pediatric Genetics and Metabolism
The University of North Carolina at Chapel Hill

Susan A. Berry, M.D.
Professor and Director
Division of Genetics and Metabolism
Departments of Pediatrics and Genetics,
Cell Biology & Development
University of Minnesota

Jeffrey P. Brosco, M.D., Ph.D.
Professor of Clinical Pediatrics
University of Miami School of Medicine
Department of Pediatrics
Deputy Secretary, Children's Medical Services
Florida State Department of Health

Kyle Brothers, M.D., Ph.D.
Endowed Chair of Pediatric Clinical and
Translational Research
Associate Professor of Pediatrics
University of Louisville School of Medicine

Jane M. DeLuca, Ph.D., R.N.
Associate Professor
Clemson University School of Nursing

Scott M. Shone, Ph.D., HCLD(ABB)
Director
North Carolina State Laboratory of Public Health

EX-OFFICIO MEMBERS
Agency for Healthcare Research & Quality
Kamila B. Mistry, Ph.D., M.P.H.
Senior Advisor
Child Health and Quality Improvement

Centers for Disease Control & Prevention

Carla Cuthbert, Ph.D.

Chief, Newborn Screening and Molecular Biology Branch

Division of Laboratory Sciences

National Center for Environmental Health

Food and Drug Administration

Kellie B. Kelm, Ph.D.

Deputy Director

Division of Chemistry and Toxicology Devices

Office of In Vitro Diagnostics and Radiological Health

Health Resources and Services Administration

Joan Scott

Director

Division of Services for Children with Special Health Needs

DESIGNATED FEDERAL OFFICIAL

Catharine Riley, Ph.D., M.P.H.
Health Resources and Services Administration
Genetic Services Branch
Maternal and Child Health Bureau

ORGANIZATIONAL REPRESENTATIVES

American Academy of Family Physicians
Robert Ostrander, M.D.
Valley View Family Practice

American Academy of Pediatrics
Debra Freedenberg, M.D., Ph.D.
Medical Director, Newborn Screening and
Genetics, Community Health Improvement
Texas Department of State Health Services

American College of Medical Genetics & Genomics
Michael S. Watson, Ph.D., FACMG
Executive Director

Association of Maternal & Child Health Programs
Jed L. Miller, M.D., M.P.H.
Director, Office for Genetics and People with Special Health Care Needs
Maryland Department of Health
Prevention & Health Promotion Administration

Association of Public Health Laboratories
Susan M. Tanksley, Ph.D.
Manager, Laboratory Operations Unit
Texas Department of State Health Services

Association of State & Territorial Health Officials
Christopher Kus, M.D., M.P.H.
Associate Medical Director
Division of Family Health
New York State Department of Health

Association of Women's Health, Obstetrics, & Neonatal Nurses
Jacqueline Rychnovsky, Ph.D., R.N., CPNP, FAANP

Vice President, Research, Policy, and Strategic Initiatives

Child Neurology Society

Jennifer M. Kwon, M.D., M.P.H., FAAN
Director, Pediatric Neuromuscular Program
American Family Children's Hospital
Professor of Child Neurology
University of Wisconsin School of Medicine & Public Health

Department of Defense

Jacob Hogue, M.D.
Lieutenant Colonel, Medical Corps, US Army
Chief, Genetics
Madigan Army Medical Center

Genetic Alliance

Natasha F. Bonhomme
Vice President of Strategic Development

March of Dimes

Siobhan Dolan, M.D., M.P.H.

Professor and Vice Chair for Research

Department of Obstetrics & Gynecology and

Women's Health

Albert Einstein College of Medicine and Montefiore

Medical Center

National Society of Genetic Counselors

Cate Walsh Vockley, M.S., LCGC

Senior Genetic Counselor

Division of Medical Genetics

UPMC Children's Hospital of Pittsburgh

Society for Inherited Metabolic Disorders

Georgianne Arnold, M.D.

Clinical Research Director, Division of Medical

Genetics

UPMC Children's Hospital of Pittsburgh

C O N T E N T S

P R O C E D I N G S

DR. CYNTHIA POWELL: Good morning, everyone. Since we're meeting by webinar today, I wanted to introduce myself. I'm Cindy Powell, Chair of the Advisory Committee on Heritable Disorders in Newborns and Children. I would like to welcome you to the Committee's fourth meeting in 2019. We will begin by taking the official roll call. Kamila Mistry.

DR. KAMILA MISTRY: Here.

DR. CYNTHIA POWELL: Mei Baker. She's unavailable. Susan Berry.

DR. SUSAN BERRY: I'm here.

DR. CYNTHIA POWELL: Jeff Brosco.

DR. JEFF BROSCO: I'm here.

DR. CYNTHIA POWELL: Kyle Brothers. I believe he'll be joining us later this morning. Jane DeLuca.

DR. JANE DELUCA: Here. Scott Grosse.

DR. SCOTT GROSS: Here.

DR. CYNTHIA POWELL: Kellie Kelm.

DR. KELLIE KELM: Here.

DR. CYNTHIA POWELL: Joan Scott.

JOAN SCOTT: Here.

DR. CYNTHIA POWELL: I'm here. Diana Bianchi. I don't she's available.

DR. CYNTHIA POWELL: Annamarie Saarinen. I don't think she was going to be available today. Scott Shone.

DR. SCOTT SHONE: Here.

DR. CYNTHIA POWELL: Beth Tarini. Catharine Riley.

DR. CATHARINE RILEY: Here.

DR. CYNTHIA POWELL: And now, we'll go through those attending from the organizational representative group. Robert Ostrander.

DR. ROBERT OSTRANDER: Here.

DR. CYNTHIA POWELL: Debra Freedenberg.

DR. DEBRA FREEDENBERG: Here.

DR. CYNTHIA POWELL: Mike Watson.

DR. MICHAEL WATSON: Here.

DR. CYNTHIA POWELL: Steven Ralston. Jed Miller.

DR. JED MILLER: Here.

DR. CYNTHIA POWELL: Susan Tanksley.

MS. SUSAN TANKSKLEY: Here.

DR. CYNTHIA POWELL: Chris Kus.

DR. CHRISTOPHER KUS: Here.

DR. CYNTHIA POWELL: Jacqueline Rychnovsky.

DR. JACQUELINE RYCHNOVSKY: Here. DR. CYNTHIA POWELL: Jennifer Kwon. Jacob Hogue.

DR. JACOB HOGUE: Here.

DR. CYNTHIA POWELL: Natasha Bonhomme.

MS. NATASHA BONHOMME: Here.

DR. CYNTHIA POWELL: Siobhan Dolan.

DR. SIOBHAN DOLAN: Here.

DR. CYNTHIA POWELL: Cate Walsh Vockley.

MS. CATE WALSH VOCKLEY: Here.

DR. CYNTHIA POWELL: Georgianne Arnold.

DR. GEORGIANNE ARNOLD: Here.

DR. CYNTHIA POWELL: All right. Now, we'll look at the August minutes. The Committee members received a draft of the August meeting minutes to review prior to this meeting. No edits

were submitted. The Committee received the final draft of the minutes prior to the meeting. Are there any additions or corrects to the minutes before we take a vote? Okay. None being heard, we'll go through those available to vote on the August minutes. Mei is not available. Susan Berry.

DR. SUSAN BERRY: Approved.

DR. CYNTHIA POWELL: Jeff Brosco.

DR. JEFF BROSCO: I missed that. Are you asking for a vote?

DR. CYNTHIA POWELL: Yes, state either yes, no, or abstain in terms of approving the August 2019 minutes.

DR. JEFF BROSCO: Okay. I abstain -- I abstain, as I was not present.

DR. CYNTHIA POWELL: Kyle Brothers, I don't believe is available yet. Jane DeLuca.

DR. CATHARINE RILEY: I'm sorry, this is Catharine. Jane is trying to log in. She's having difficulties.

DR. CYNTHIA POWELL: Okay. All right.

Scott Grosse.

DR. SCOTT GROSSE: Approve.

DR. CYNTHIA POWELL: Kellie Kelm.

DR. KELLIE KELM: Approve.

DR. CYNTHIA POWELL: Kamila Mistry.

DR. KAMILA MISTRY: Approve.

DR. CYNTHIA POWELL: I approve. Scott Shone.

DR. SCOTT SHONE: Approve.

DR. CYNTHIA POWELL: And Joan Scott.

MS. JOAN SCOTT: Approve.

DR. CYNTHIA POWELL: Anyone on the Committee who is available whose name I didn't call? Okay. All right.

At the August meeting, I introduced new organization representatives including Lieutenant Colonel Hogue representing the Department of Defense. Dr. Hogue is the Chief of Genetics at Madigan Army Medical Center located on Joint Base Lewis-McChord in Tacoma, Washington. Dr. Hogue was not able to join us in August but is with us on the webinar today, and we would like to welcome

Dr. Hogue. Thank you for serving.

Next, I wanted to provide an update on the medical foods report, which the Committee previously accepted. An informational copy was sent to the Secretary. On September 9th, HRSA's Acting Administrator sent a reply on behalf of Health and Human Services thanking the Committee for providing an informative summary of the current landscape of medical foods in the United States and outlining the challenges faced by individuals living with inborn errors of metabolism.

So, our meeting topics for today include Data Interoperability in Newborn Screening, which we started taking a look at at our last meeting and then to continue with the review of the RUSP Condition Nomination and Evidence Review Process, specifically today to look at the Public Health System Impact Assessment.

At this time, I would like to turn things over to Joan Scott from HRSA.

MS. JOAN SCOTT: Good morning, everyone.

This is Joan Scott from HRSA, and I just wanted to make a quick comment before Catharine got on the line to do her DFO comments.

So, as you know, the legislative authority for this Committee is set to expire on September 30th. At that time, if the authorizing legislation has not passed, Committee operations will halt. However, operations for continuing the work of the Committee are being considered. As you know, the legislation -- current legislation does provide for the option of establishing a discretionary committee, and that is one of the options that is being considered. So, just watch the Committee website for information and all of the future dates remain on the Committee's website. They are tentative pending what will happen, but just watch the Committee's website for further information. Thank you, and now I'll turn it over to Catharine.

DR. CATHARINE RILEY: Thank you, Joan. We're just going to pause here for a minute while we're trying to get the slides loaded so everyone

on the webinar can see them. So, while we're loading the slides, I'll just go ahead and get started. This is Catharine Riley. I'm the Designated Federal Official for the Advisory Committee on Heritable Disorders in Newborns and Children. I first just want to say welcome to everyone who is joining us on this webinar from all across the US. We know there are folks joining from different time zones, so especially those on the west coast and those early time zones, thank you for joining us early this morning.

This Advisory Committee legislative authority is found in the Newborn Screening Saves Lives Reauthorization Act of 2014. This legislation established the Committee and provides the duties and scope of work for the Committee. However, all community activities are governed by the Federal Advisory Committee Act or FACA, which sets the standards for the establishment, utilization, and management of all Federal Advisory Committees. As a Committee member on a

Federal Advisory Committee, you are subject to the rules and regulations for special government employees.

I also have standard reminders to the Committee that I would like to go over. I wanted to remind the Committee members that as a Committee, you are advisory to the Secretary of Health and Human Services, not the Congress. For anyone associated with the Committee or due your membership on the Committee, if you receive inquiries about the Committee, please let Dr. Powell and I know prior to committing to an interview.

I also wanted to 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 received a waiver from HHS authorizing you to participate.

When a vote is scheduled or an activity

is proposed and you have a question about a potential conflict, please notify me immediately.

I'll pause here and see if any Committee members have any questions in regard to conflict of interest. Okay. Next slide please.

So all Committee meetings are open to the public. If the public wishes to participate in the discussion, the procedures for doing so are published in the Federal Register Notice and are announced at the meeting. So, for this meeting, the request we were offered two options, an oral -- provide oral comments or provide written comments. We did receive two requests for oral comments, so we'll share those later this morning, and we did receive one written statement ahead of time, and the Committee members were provided a copy of that written statement prior to the meeting.

Any further public participation will be solely at the discretion of the Chair and myself as DFO.

Any questions, again, from Committee

members before proceeding? Okay. Next slide.

I just wanted to, since we are on a webinar, just go over some of the instructions and to thank everyone for their patience this morning, as we were dealing with some technical difficulties, and we're continuing to work through those. So, thank everyone for your patience.

For members of the public, the audio will be coming through your computer speaker. So, there is a call-in option, and you can listen through a phone as well, and that number is listed on the screen. But you should be able to hear through your computer to the webinar.

For Committee members and organizational representatives, your audio will be coming through the phone line that you called in on, so if you could please make sure you have your computer speakers turned off, this will help when you are providing comments or questions so there won't be an echo.

I'm asking Committee members and organizational representatives to please speak

clearly and remember to state your name first to ensure proper recording for the Committee transcript and minutes. If you are having any issues with your phone line, you can press star zero to reach the operator.

In order to facilitate the discussion, please use the raise hand feature in the Adobe Connect when you are wanting to make comments or ask questions. At the top of your screen, you'll see a little raise hand. I will see this, and as soon as I have that noted, we'll make a list, and I will clear that. So, if you see that cleared, we know you haven't made a comment, we're just trying to keep the list running. So, if it's cleared, that means we've seen it and you're on the list and your in the queue for providing a comment or asking a question. If you're having any trouble with that for Committee members, you can E-mail me directly at Catharine.

And if you're having technical difficulties or if the webinar pauses during it, if you could try to close out and reopen the

webinar using a different browser, that can help. If you're still having technical difficulties, please refer to the contact information provided in the registration confirmation E-mail that you received, and they will be able to help troubleshoot that.

So, with that, I am going to turn it back over to Dr. Powell. Thank you.

DR. CYNTHIA POWELL: Thank you, Catharine. At the August meeting, we heard an excellent overview of Data Interoperability in Newborn Screening. We heard about the differences between data exchange, data interfacing, and data interoperability. We also heard about some aspects of newborn screening that could benefit from the use of interoperability through databases including specimen tracking, electronic orders and reporting, which we will hear more about today, hearing and critical congenital heart disease screening, record and birth defect registries, which we'll hear more about today, long-term followup, pediatric specialty care, and

immunizations.

Today, we'll hear from two states, Texas and Minnesota, about their experience implementing electronic test ordering and automatic daily electronic data transfers between vital records and newborn screening. Brendan Reilly will share the Texas experience and Amy Gaviglio will share the Minnesota experience. We will hear from both presenters and then open it up for questions and discussion. We hope to hear from additional states at future meetings.

I'd like to introduce our first speaker. Brendan Reilly is a Program Specialist for the Texas Department of State Health Services Laboratory. He has over 18 years of experience managing projects related to quality improvement, process workflow, and informatics. He is co-Chair of the Newborn Screening Technical Assistance and Evaluation Program known as NewSTEPs Steering Committee, co-Chair of the Newborn Screening Health Information Technology Work Group, and Moderator of the Newborn Screening Health

Information Technology Interoperability User Group.

DR. CATHARINE REILLY: Brendan Reilly, are you on the line? If you are, we're going to give you a minute to respond. Operator, if Brendan Reilly is on the line but doesn't have an open line, can you please move him to an open line?

DR. CYNTHIA POWELL: Due to some technical challenges at the moment, I'm sorry, Brendan, we're going to have Amy Gaviglio go first. So, I'll introduce Amy.

Amy Gaviglio, a Certified Genetic Counselor, worked for the Minnesota Department of Health Newborn Screening Program for the past 12 years, where she oversaw followup and provided guidance for informatics, education, ethical and policy-related initiatives. Ms. Gaviglio is in the process of transitioning to a new position within the newborn screening community. She is a member of the Committee's Education and Training Workgroup and the Ad Hoc Workgroup on interpreting

newborn screening results. She also currently serves as co-Chair of APHL's New Disorder Work Group and is a member of APHL's Short-Term Followup and Legal Legislative Issues in Newborn Screening Work Group.

Amy, hopefully you're on, and you're going to be talking about building connections to improve outcome.

MS. AMY GAVIGLIO: Thank you, Dr. Powell. Can you hear me?

DR. CYNTHIA POWELL: Yes, we can hear you, Amy.

INTEROPERABILITY FOR NEWBORN SCREENING: STATE EXPERIENCES

MS. AMY GAVIGLIO: Oh, perfect. Okay. So, thank you too, Dr. Powell, as well as the Committee for allowing me the opportunity today to speak about how building data can actually really help improve newborn screening outcomes. Next slide, please.

So, I'd like to start with a quick disclaimer that some of the work presented here

was completed during my tenure at the Minnesota Department of Health. As Dr. Powell mentioned in my bio, I'm currently in the process of transitioning positions, so I'm no longer an MDH employee. Next slide.

So, when talking about a rather broad and somewhat conceptual topic like interoperability, I like to start with a why and why might we as a newborn screening community or why might a newborn screening program want to take the time and effort toward building more data connections. And the answer to this really spans the entirety of the newborn screening process. From the preanalytical perspective, electronic connections can improve upon the integrity of the data coming into the program, which ultimately can improve upon the result accuracy.

In addition, connections can aid in providing a more accurate denominator within the state, and this, of course, will help allow for the assessment of refusals.

Analytically, having accurate demographic

information can help ensure that the appropriate age or birth weight-based cutoffs are applied or that low birth weight infants are screened per LBW serial screening recommendation. This, of course, not only aids in better screening but can also ensure that delays don't occur while waiting for correct demographic information.

Certainly, we think from a point-of-care perspective, so [inaudible] and PCHD, electronic connection really can be pivotal in greatly improving the program's ability to receive and monitor the screening results themselves.

In the post-analytical sphere, better data connections may reduce reporting errors, they can help staff locate infants who have actionable newborn screening results, as well as assess the long-term outcomes of families identified through newborn screening in the hopes of being able to answer the question of whether we are truly meeting the mission of newborn screening.

For my time today, I will be focusing on -- if you can do the next slide -- these two

components, so the accurate denominator -- next slide -- as well as the assessment of outcomes, and Brendan will cover some of the others in his talk. Next slide, please.

So, a key tenant of newborn screening is that every newborn should be given the opportunity to have newborn screening. However, not surprisingly, it can be rather difficult to see that through if you do not have a way of knowing how many births there are -- of having an accurate denominator. And this is where connecting to vital records can really come into play. So, by matching specimens received to birth certificates filed, one can better know who has and hasn't been screened, follow up accordingly, and also monitor refusal rates.

So, before talking more about how this can work, I want to point out three areas of consideration for creating this linkage. The first is really the importance of understanding the statutory requirements in each state regarding birth certificate filing, particularly any

stipulations around required timing. So, how long do birth attendants have to file the birth certificate, and is this timing different for midwifery or out-of-hospital births.

The second and somewhat related to the first is to understand the limitations that may still exist in linking to vital records, both in terms of timing -- so, for example, if certain populations take longer than a month or so to file their birth certificates or if there are populations that may still want to be screened, but their home birth certificates may not be filed for months, years, or maybe never, so it's important to understand that these may still not be captured by the linkage.

And really, both of these come together for the last point, which is that really in order for linking to vital records to truly help in moving the mission of newborn screening, it really needs to be done in a timeframe that allows program intervention if a child is inadvertently missed. Next slide.

So, how can this be achieved? So, this slide illustrates the process that has been utilized in the Minnesota program since August 2016. And this process starts with our Office of Vital Records sending a daily file. You can just kind of like tab through the next slide for the animation. The Office of Vital Records sends a daily file of birth certificates filed the previous day. This is sent through our Internal Exchange Hub and ends up in a .csv file on our network. At this point, a Newborn Screening Program staff person kind of manually goes, grabs that csv file, and imports it into our LIMS, and this happens each day. Next slide.

And again, if you just want to tab through, thank you. So, once imported, a query is run within our LIMS that looks to match the birth certificate information with specimens received. And so, based on some probabilistic matching analysis that was done, we found that the four criteria of infant's date of birth, infant's time of birth, and mother's first and last name gave

the most automatic matches with the highest accuracy. So, upon running that query, if a match is obtained using that criteria -- those four criteria points -- the birth certificate number and any associated information is automatically added to the patient's case within the LIMS. If a match is not obtained, the remaining records are then manually reviewed by program staff, who deselect or select other demographic criteria to try to determine if a match exists. For example, time of birth -- that can often be off by one or two minutes, so that may be preventing a match from occurring. So, if you deselect that, you may find a match.

Again, during this process, if a match is determined, then a birth certificate number and information can be added to the case at the click of a button. If no match is obtained after about five to seven days, there is no refusal paperwork, or there has been no notification of a deceased status, then followup can begin to determine why a specimen has not yet been received. Next slide.

Using this process, we are able or the Minnesota program is able to perform a match in just over five days from date of birth and then, of course, more shortly after the date the birth certificate is filed. The significance of these numbers is that this timeline allows the program the potential to intervene if a child has been missed before most symptoms might -- might occur. This process also allows the program to capture a number of different things, so number of screening refusals, who has been born in this state, transferred out but still ensuring that that screen occurred, how many infants had specimens that were lost and needed followup to get a new specimen, as well as how many infants remained unscreened even after multiple contacts. Next slide.

In addition to connecting with vital records, there is another opportunity for obtaining a state-level denominator at least from birth facilities with EMRs, and this is known as Newborn Admission Notification Information or

NANI. So, this process connects to the birth facility's EMR using an HLT ADT feed, which is a different type of connection than what Brendan will describe in his talk. In Minnesota's implementation of this with our systems, four specific ADT messages are received from birth facilities. The first is called an AL1 message, and this actually alerts the program that a child has been born or admitted. This is then followed by several other messages -- several AO8 messages. So, those messages update the patient's record as information is added. For example, once the birth weight is added, a new message would be sent with parent contact information, et cetera. And then, once the child is discharged, a third type of message called an AO3 is sent. A fourth type of messages called an A31 is also very helpful, particularly for getting the child's legal name, as we found in many facilities that this is actually not updated until after the child is discharged. So, you can see that, you know, from getting these four different messages types, one

can not only know when a child was born -- so, again, kind of helping with that denominator -- but also can know if the child is still inpatient, so in the hospital, and this can be really helpful for followup staff as they are calling out results to know kind of where the baby might be. However, of course, just like kind of the issue with vital records and actually even more so here, this will not accurately account for out-of-hospital births. Next slide.

All right. And then again, you can tab through to the animation. Thank you. So, this is what a NANI connection looks like. In this case, we're starting with the birth facility and their EMR where the HL7 ADT feed is set up to filter the newborns, so we only want messages on individuals less than 72 hours of age and then those -- it will also send all of those subsequent messages. These messages eventually end up going through the Internal Exchange Hub again, and they are then placed in a holding table in the LIMS.

Simultaneously or kind of shortly after

the specimen is collected at the birth facility, couriered to the Public Health Lab, where upon receipt, the bar code is scanned, a few demographics entered, and then the message and the specimen are matched together. Next slide, please.

So, I think you will see somewhat of a similar timeline in Brendan's talk as well, but projects involving connecting to birth facilities are really not for the faint of heart. It can take quite a long time. Minnesota implementation began in 2014 and concluded with all approximately 90 birth hospitals being connected in early 2017.

Two key points here I'd like to touch on include in 2015 the declaration of this reporting mechanism as a public health registry for the purpose of meaningful use, now known as promoting interoperability, and this really greatly helped incentivizing hospitals to work with the program on this project. And then the second point is that, you know, much like any other project, once implementation concludes, you really enter a new

phase of continuous monitoring to ensure that the feeds continue to work, the demographics look the way you expect them to look, and then adjusting any work flows that you need to with this new process. Next slide.

So, as Dr. Powell mentioned kind of in her introduction, vital records and NANI, are just two of many possible connections a program may make in order to help improve outcome. Some of these are internal connections. They're internal in terms of interagency, as is the case for birth defects. So, this connection can be really important in helping programs ascertain missed cases. That has been especially true to critical congenital heart disease. But, they may also help better understand co-morbidities present in newborn screening conditions and maybe considerations that need to be made around screening in that population. So, an example of this can be infants with Down syndrome who also have congenital hypothyroidism.

Additionally, connecting to other

programs like WIC, children with special health needs, or local public health can also aid in followup, particularly ensuring access to care and treatment.

Externally, there are opportunities for connections as well, of course, so connections to reference or clinical laboratories and subspecialist EMRs have the great potential to improve our ability to obtain followup information both in the short term, but I think more importantly in the long term to better ascertain things like change in state or clinical status for late-onset cases, the ability to be alerted to new diagnostic or outcome information, or to, of course, false-negative cases. Next slide.

So, I borrowed this slide -- this graphic from the Office of the National Coordinator for HIT, as I think it does a really great job of reminding us of how this work has the potential to not only aid in improved individual-level outcomes but shows how data connections and interoperability more broadly can impact the

population level by informing our understanding of disease, by shaping the clinical guidelines, and effecting public policy. This paradigm is, I think, even more important in the world of newborn screening in rare disease as really, I think, only through more robust data connections and improved data integrity will we be able to truly affect the impact of newborn screening and inform new and best practices. Next slide.

So, how do we proceed? How do we get there? First, I think we must understand that programs are being asked to do more things more quickly with more complexity and really have limited resources that are pulled in any number of directions at any given time, and certainly -- and I think you'll see this with Brendan's talk -- interoperability has the potential to help by saving staff time and money and perhaps even improving the newborn screening process and system. But the catch 22 is that in order to make it happen, you need the time and money upfront, which ultimately means that the starting point for

each program kind of on their journey toward interoperability is likely to be different and really depend upon their own needs assessment, gap analysis, and really ability to add this to their priorities. Next slide.

You can tab through, thank you. So, I wanted to end with this quote from Karen DeSalvo at the Department of Health and Human Services, because I think this really describes exactly what our next steps and goals should be in newborn screening, and that is to create a road map with programs to achieve interoperability while really keeping the why -- the ultimate mission of newborn screening in mind. But, before we all embark on this journey, programs will need help packing their bags, so to speak, so you can tab through again a little bit. So, in terms of what help is needed, of course, nationally coordinated efforts in providing technical assistance, training fellowships, standards and national initiatives, as well as, of course, funding. So, next slide, please.

So, with that, I would like to acknowledge the great team at the Minnesota Department of Health and thank you again for the chance to speak today, and I will pass it over to Brendan.

DR. CATHARINE RILEY: Hi, this is Catharine Riley, DFO. We wanted to ensure -- Brendan Reilly, are you on the line? Okay. We are having technical difficulties. We know Brendan Reilly is on the line, but we can't hear him. So, Brendan, if you can hear us, we cannot hear you at this point. If you could push star zero and let the operator know that you need an open line, please.

DR. CYNTHIA POWELL: Hi, this is Cindy Powell. Thank you very much, Amy. I think as we work out our technical difficulty being able to hear Brendan Reilly's presentation, we'll go ahead and take questions from first Committee members and then organizational representatives. So, operator, if you can please open the lines for Committee members and organizational reps. We'll

have Committee members ask their questions first, and as a reminder, please use the raise hand feature in Adobe Connect when you would like to make a comment or ask a question, and as always, please state your first and last name each time you ask a question or provide comment in order to ensure proper recording.

MR. BRENDAN REILLY: Hi everybody. This is Brendan Reilly. I think I may have finally gotten access.

DR. CYNTHIA POWELL: All right. Before we lose you, Brendan, let's hold off on questions then, and we'll go ahead and Brenda, if you can give your presentation on newborn screening in Texas.

MR. BRENDAN REILLY: Yes.

DR. CATHARINE RILEY: Brendan, I'm sorry, this is Catharine, and for the record, I wanted to note we do have Kyle Brothers and Jane DeLuca, Committee members, who have joined the webinar. Thank you.

MR. BRENDAN REILLY: All right. Thank

you, guys, and I apologize for the difficulties.

DR. CYNTHIA POWELL: Oh, no problem.

MR. BRENDAN REILLY: It's interesting to have a presentation on technology when technology can be a little difficult at times. So, with that said, let me move forward with the presentation, and thank you for your patience with me, everybody.

So, I am going to speak on some interoperability efforts that we have taken in Texas over the last ten years and to set that up, I wanted to review this slide that was presented by Ashleigh Ragsdale at the last Committee meeting to kind of give an overview and this slide gives an overview of the different activities that we do in newborn screening programs for dried blood spot testing and followup. The functionality in blue is activities that are -- that we use -- that we currently do and use our information systems to manage, and those activities in orange are some of those activities that we could see as future functionality with the proper interoperability

infrastructure and design, and, you know, I -- Amy already was able to give a presentation on some of that functionality and so that's just the context in which I want to talk about what we're doing for electronic test ordering and reporting and so if you see the green circles here, this is the functionality that I'm focusing on. This is an interoperability functionality, but it's something that we've been doing here in Texas for a while and working on pretty -- pretty extensively.

Generally, over the last ten years when the community has discussed interoperability, this is what we're talking about -- specifically electronically test ordering and reporting. So, I kind of want to -- the idea is to set this up as kind of a historical perspective on that with the understanding that there's all kinds of other functionality out there that we're capable of achieving.

So, I am going to talk about electronic test orders. Traditionally in newborn screening programs, these are received on a demographic form

that comes along with the blood spot specimen, and all the information is handwritten onto the form. We're limited to the number of fields that can be collected and fit on that 4-1/2 by 11 form or whatever it is -- whatever the size is. And generally, the information is hand transcribed onto it, received by data entry operators, and hand entered into a system separately where you could have all sorts of transcription issues.

So, the way that this works in electronic order is obviously this is going to be sent electronically, but it's a little bit more intricate than that in general. So, usually the order for the newborn screening is placed in the electronic medical record of the hospital system by the physician or by a standing order or something like that, and then there's a couple of ways that that information in most cases is actually transmitted to a different information system within the hospital. So, the order message may be transmitted to their laboratory information system or as Amy was describing with the NANI

system, an admission message can be sent to a web application or to potentially a laboratory information system. And then, laboratory -- generally, laboratory staff or phlebotomists will access that laboratory information system for a newborn screening specimen and transmit a separate order that will go out to the public health laboratory through integration engines. So, an integration engine takes the information, puts it in a proper format, and sends it on to the public health laboratory.

So, that's a nuance to it, but it's an important nuance when we start looking at the actual data flow and how things usually operate for reference laboratory testing in a public health laboratory, and I'll come back to that in a little bit.

On the other end of things, when we're talking about electronic results, you know, traditionally and historically, we're sending -- generating a physical result report with all the results for newborn screening sample that are

mailed to a provider and then somebody on that end will take all that information and transcribe it to the hospital information system -- the EMR through a similar pathway. And here, you can see in the electronic result, it's electronically transmitted. It shows up into the system without any transcription and with different coding that you know, the system can use to automatically flag things and things of that nature.

So, a little history. Many of you may have already seen some of this background for Texas, but this is an updated version. So, we initiated our HL7 solution and implementing a web application with our vendor way back in 2008, and then we immediately started on four different projects or implementation with various hospital systems, and ultimately, through these four projects, we implemented bidirectional HL7 messaging with four partners -- that equates to forty-one facilities. This was prior to the implementation of the PHII NLM implementations guides, so the Public Health Informatics Institute

worked with the National Library of Medicine and, I believe, under the eye of this Committee, to develop an implementation guide for how to structure HL7 orders and results. So, that said, this guidance came out after our solution was put into place. Our solution was on HL7 Version 2.3, and this guidance follows HL7 Version 2.5. and without going into the details, you'll just have to trust me that there's pretty significant differences between those two HL7 standards.

So, over the next few years, you can imagine, there are lots of things going on and lots of competing projects in the newborn screening program to expansion of screening tests, upgrade of information systems, you know, RFPs for services, all sort of competing issues essentially slowed down the process. Then, it came down the road that the implementation guides -- the PHII NLM implementation guide, there was an effort led to actually align this -- these implementation guides with the HL7 standarized laboratory orders interface and laboratory results interface guides,

and knowing that that was coming, we, you know, started developing implementation of our capability to receive those inbound orders, which we did and went live with in 2017, and our partner in this case was OZ Systems, who is providing these services to about fifteen facilities for us right now and currently, that is just an inbound orders interface.

And so, following that, we also revised our electronic reporting to match the Version 2.5 implementation guides and followed the same standards that other states are working on as well, and we just recently here in this last August implemented with transitioning one of our original partners over to this new system, and they have gone live -- they have been live with us for about a month. And we've also now initiated four additional projects to transition existing facilities and also add new facilities and add the results piece with -- with OZ Systems. So, a lot of extensive effort going on in that space and ultimately our goal is that once this is done,

we'll have somewhere around eighty facilities that are participating in our bidirectional interface.

So, what does that look like in terms of successes? The obvious goal here is to try to achieve quality improvement and efficiency improvement on both sides of things. So, we were able to track some data -- some hard data for improvement to our system. But keep in mind there's efficiencies gained on both sides, as I kind of review some of these.

So, ultimately at our current rate, we're receiving about 130,000 electronic orders per year either through an automated electronic message or through web application that we have available. It only accounts for 17 percent of our samples, which doesn't sound much, but with our volume, that's 130,000 samples a year. And on the reporting end of things, we're reporting about 117,000 results via electronic HL7 message.

So, what does that equate to in terms of some program efficiencies? For the order entry capability, we've determined and estimated that it

takes about two minutes per sample to manually key in all of the information, and when we extrapolate that out to the number of FTE per year, we're estimating that's close to three FTEs per year, and that's at our current rate, and we -- we're working on solutions to further advance that with some of the additional projects that we're working on now.

In terms of data accuracy, we were able to do a small study, where we actually received specimens with not handwritten information but a label that was generated out of the system that was transmitting the electronic order. So, the label information in this case, we knew exactly matched what was received in the electronic order, and in this case, we were only receiving the electronic order into our test system during a validation process, so -- and the information on the label was being entered by our data entry operators. So, we were able to compare the two data sets to look for differences in the data sets. Now, it's important to note that there are

some allowed differences from what's on the form by our data entry operators, so that could account for some of these differences, and I'd also add that we have a very extensive quality assurance process for our data entry operators and an experienced team that really does a huge amount of work to get this information entered. But the best team in the world is still being asked to do this in a crazy fast amount of time, you know, and I think this data kind of backs up that, you know, even their most robust efforts to get this data entered accurately is not going to match the efficiency of an electronic data transfer.

In terms of data completeness, we're also able to measure hospital systems that implemented electronic ordering and track the number of missing key data elements before and after implementation and I think this pretty well outlines some of the improvement that we saw for some of these key data elements being included, which again represents a really huge timesaving in terms of getting the sample run and not spending

time and resources following up to try to get that key information and insuring that our testing is based on the most robust data quality.

And in this example I'm showing -- this is actually an HL7 provider -- and we can -- you can see here the percent of samples where we receive all the key data elements that we're requesting as opposed to the state average for that -- for that measure.

So, before I move on, that's just a reminder that's just a few of the benefits that we see on the public health side of the program, but please keep in mind that, you know, a lot of these efficiencies are also occurring on the hospital side and, you know, so we're not tracking all of that.

That said, there are challenges, as Amy mentioned, and my technological difficulties today kind of demonstrate, that these things aren't always as easy as hey, just set it up and get it going. And one of the most common questions I get in terms of the challenges of advancing electronic

testing or reporting is how do we get and keep healthcare providers engaged in this process? And my response to that is generally that it's not my belief that hospitals and healthcare providers in general are not interested in this, they are certainly interested in it, and I feel like they, you know, there's a big push from hospital systems to gain these efficiencies. But the problem is that it's not always quite as easy an effort once they start looking into it.

And so, let me talk about some of the deficiencies of the model that we've implemented here in Texas or at least some of the difficulties that we run into.

So, here's another slide that Ashleigh presented in her great presentation last month that kind of lays out the ideal perspective, at least in my mind, for how a hospital would like to see interoperability work, and you can see in this case, the hospital has one connection to an agency. We're in the laboratory up here in this little left-hand corner of the laboratory box, but

there are all kinds of different agencies requesting information from hospitals and trying to -- to share that information from the hospital. So, this would be the ideal solution. Here in Texas, our solution looks a little bit more like this. So, we're setting up direct interfaces with each of those hospitals and then if the hospitals want to communicate with the other parts of the agency, then they would be required to set up a separate connection with each of those. So, that's -- that's one deficiency that we -- that we have, and it's also a hurdle that the hospitals have to jump each time they have to work on getting that one more connection set up.

So, another issue that we've run into quite a few times when we're working with the hospitals is going back to the slide for how the test order works. So, this is a standard, I think, you know, this is my understanding of how a standard reference laboratory test order works at a hospital for something like, I don't know, a glucose panel or -- or something of that nature.

So, in the EMR, the healthcare provider will place the test order, and the information that's passed to their laboratory information system is the information on that patient and specifics as to what's being ordered. So, I'm ordering a glucose panel. Now, obviously, most hospitals will probably be able to run this in-house, but if it's a test ordered that needs to be sent out to a reference laboratory, they'll send that information out of their laboratory information system. It will include that patient information and test order information that they received from the EMR, and then they'll add the specimen level information for what's collected, and it will be sent out to that reference laboratory.

So, how it works a little bit differently in newborn screening, and this is significant to note that newborn screening is different from a standard reference lab test order. So, in newborn screening, the EMR will again send the patient -- or in this case, it's actually not a patient, it's a newborn -- and their test order information to

the LIS, and then the LIS system is being asked to then transmit that newborn information -- which I'll reiterate is different from, you know, my personal patient information if I'm having a test order placed -- it will send the test order and specimen information, but we're -- newborn screening programs are also asking for all the mother's information and that magical post-discharge provider information or who the PCP will be for that provider for that newborn after they've been discharged from the hospital. So, all this information generally is not available in the laboratory information system, and the providers -- the partners will have to scramble to figure out how to do custom coding to get all that information into the message going to the laboratory as the majority of LIS and EMR vendors don't necessarily have specific solutions just for newborn screening. They'll have a standard reference lab solution and then additional work will have to be done to get the newborn screening information in.

A few other pain points, just touching on them quickly -- and again, this is a non-exhaustive list of some of the challenges to this -- generally, we're finding that hospitals are having some issues with receiving multiple disorder results for a single panel order. So, if you think about it this way, they place an order for a newborn screening panel, and when we send results back, we're sending results back for a list of orders for amino acid disorders, fatty acid disorders, CAH, et cetera, and so this is something that we've been kind of working through with the hospital to figure out how is best to do it, and unfortunately, different hospitals want to do it different ways.

So, another thing -- and this is a little bit difficult to describe -- is some of the variation in what I would call adherence to standards. We've been very strict about our approach in exactly following the standard, but that said, we do leave our solution open to where we follow the standard, but if our partners don't

want to follow it to the tee, we leave some flexibility for them. But, that said, as we're -- we are running into situations where we're asking partners to -- or where our partners are asking to negotiate and change our standard ordering and do things differently just for them that don't follow the standard. And to date, we've resisted that and are not doing so, and generally there are solutions in there, but at least my impression is that often, you know, they are negotiating with the reference laboratory that really just wants their business and is going to, you know, make accommodations and then ultimately we're not all following standards again. So, that's -- that's kind of one option that we're running into.

A couple other issues is that we do require a specimen labeling solution and sometimes this gives the hospital systems just as much of a headache as generating the electronic test order, and, as I mentioned before, the EMR and LIS vendors for the hospital systems are not really focused on newborn screening, and they don't have

specific solutions readily available, at least in my experience.

And then, of course, there is healthcare system priorities. They have a lot of projects going on and getting a newborn screening project on the board can be -- can take some time and can be a little bit difficult if it's not a really easy off-the-shelf solution that they can purchase from their vendor.

So, that's all that I have. I wanted to leave that with some of those challenges, because I think there is definitely some opportunity to overcome some of these challenges as a community and there are certainly some benefits that we can gain from this type of solution, and I think I've probably gone way over my time, and that's all the presentation I have. So, thank you very much for your time.

DR. CYNTHIA POWELL: Thank you, Brendan. I will now open this up for discussion, so operator, please open the lines for Committee members and organizational representatives, and

remember to use Adobe Connect raise your hand if you have a comment or question, and please state your first and last name.

OPERATOR: Thank you. We will now begin the question and answer session. If you would like to ask a question, please press star one and record your first and last name clearly when prompted. Your name is required to introduce your question. To withdraw your question, you may press star two. One moment please for our first question.

DR. CYNTHIA POWELL: So, I'll start this off. This is Cindy Powell from the Committee. I have a question for Amy. Amy, do you have an estimate as to the FTE required to take care of -- of, you know, linking with the NANI program?

MS. AMY GAVIGLIO: In terms of the implementation of it or --

DR. CYNTHIA POWELL: More of a day-to-day -- the day-to-day running of it.

MS. AMY GAVIGLIO: So, I mean, for NANI, that's really just, you know, coming over and it

gets linked upon our -- our regular data entry. So, it's really no different than normal. In fact, hopefully as time goes on and the workflow becomes more ingrained, I think it will ultimately save data entry time.

In terms of the matching to vital records, that really doesn't take very long each day. I would say it takes one person maybe a half hour to kind of go through the least daily, if that. So, it's a pretty small requirement in terms of FTE.

DR. CYNTHIA POWELL: Okay, thank you. And we'll take the first question from Scott Grosse.

DR. SCOTT GROSSE: Thank you. Brendan, great presentation. I have a question. How many of the data entry operators do you have in Texas?

MR. BRENDAN REILLY: I -- I don't know the exact number off the top of my head. But just kind of looking at the room, my guess is somewhere around fifteen-ish. So, in that ballpark, maybe fifteen to twenty data entry operators.

DR. SCOTT GROSSE: Okay. Thank you.

DR. CYNTHIA POWELL: And Kyle Brothers.

DR. KYLE BROTHERS: Yes, can you hear me?

DR. CYNTHIA POWELL: Yes.

DR. KYLE BROTHERS: Great. Actually, I just wanted to confirm that my audio was working. I don't have a question except just to say I thought this was -- both were very interesting presentations, and I really appreciate you both bringing this to us. I think this is the type of thing, you know, we need a lot more of across both the public health system and also the hospital and clinical system. So, it's just really fascinating to see how you work these things out.

DR. CYNTHIA POWELL: Scott Shone.

DR. SCOTT SHONE: This is Scott Shone. So, I don't with discretion if Brendan or Amy or both in terms of your implementation processes. So, who -- can you expand on who ends up owning the implementation and then ultimately the ongoing maintenance? Is it, you know, does this -- because, you know, my experience has been there's

often a lot of well, this is IT, no this is program, this is department. Can you talk a little bit about, you know, for those of us who are coming up later and now beginning to implement each, you know, that question is obviously for Brendan, but in general maybe even the link to vital records. Who -- who -- who in the stakeholders -- but more importantly, who is going to own this? Is it like most projects where the program has to drive a lot of this?

MR. BRENDAN REILLY: So, I'll -- I'll answer first, Amy. So, I would say that we have a really good partnership with our application support department as far as this goes in our efforts. But it is primarily, and I would say a 90 percent driven project from the program side of things. That said, you know, it's also important to remember that each one of these projects is a partnership project with a hospital system, and every one, in my experience, of these projects takes on a little bit different flavor in terms of who's the ownership of the project. We've tried

to set this up to where we set up a project to implement our solution, and then we work with partners as a consultant in their project. But we have had some partners that essentially have just said hey can we start a project and you run it. The best solution is when the hospital system has a project manager and project setup. They have your, you know, your specifications, and then we work to -- to help them out with it. I think that may have answered your question, hopefully.

DR. SCOTT SHONE: No, that's great, Brendan. Thank you. I appreciate it.

MS. AMY GAVIGLIO: This is Amy. I completely agree with what Brendan said. I do think a big part of the ownership comes from the program, and it's often initiated by the program because it's really their ask or your ask, and it then does become vitally important that you -- you do build your team. So, working with your -- your interface team within the agency, having a project manager on that side, having what you would call like a business end project manager, so someone

from the program really kind of driving that. And really, from the maintenance perspective, I do think a lot of the ownership does come from the program as well, because you're often the first to notice when something happens having that process in place and having a project manager kind of throughout the entirety post implementation as well is really important.

DR. CYNTHIA POWELL: Any questions from organizational representatives?

I have another question. This is for Brendan. I'm sorry if I missed this, Brendan, but since you're a two-screen state, how does it work with the second screen?

MR. BRENDAN REILLY: Right. So, we do receive second screens, so when I talk about the number of healthcare facilities, you know, each of our interfaces has anywhere from ten to fifteen healthcare facilities participating in it, and in -- in at least a couple of those instances, they do have their outreach clinics participating in that same interface. So, they'll use the same

information system as their other hospital and they'll transmit information through -- through the same interface and information system setup.

So, essentially when we work on an individual project, we verify that we can receive test orders from each of the participating facilities. Each facility will have identifiers that are specific to the facility and specific EHR system so we can piece those out in the electronic messaging and route things back accordingly in the results.

DR. CYNTHIA POWELL: Okay. And this is Cindy Powell again. So, currently there's probably two or three large electronic health record providers around the country, without naming names, you know, is it -- I would imagine that the hospitals that are, you know, utilizing this system have different providers for their health -- electronic health records. Is that more challenging? Are there, you know, again without naming names, are there some that are more receptive to designing things specific for the

newborn screening system?

MR. BRENDAN REILLY: Well, so I think that's a, you know, that's -- let me kind of approach that in -- in two ways. So, first you're correct. There are two or three main EMR vendors, but there's quite a few laboratory information system vendors out there, and there's really two models for how these electronic -- how the systems work in the hospital. Sometimes they'll have an integrated system to where they'll have an EMR and an LIS through the same vendor, and everything works together really sweetly and nicely. But there are a lot of cases, and many of the cases where we deal with where you'll have one EMR vendor and separate LIS vendor. So -- so, that said, our approach historically in promoting these solutions and working on them is to work with -- directly with the hospital system and in working with those hospital systems, we have had at least one of the main EMR vendors that, you know, at the request of the hospital system, modified the way their system worked. This is an integrated

system, so they had that flexibility, but they modified the standard way that their system works for all their hospital systems to where it could grab that mother's information and send it over.

But, that said, the model has been working with the hospital system and so I'm -- I personally, at least, am trying to redesign that approach and reach out more directly to some of the EMR and LIS vendors to, you know, see what we can do to advance them developing a newborn screening-specific solution that would work for any of their clients.

DR. CYNTHIA POWELL: Okay, thank you.

Is there anyone on the line who had a question or comment from the Committee members or organizational representatives in case we missed your raised hand? Okay. And before we move on, I just wanted to give Kyle Brothers and Jane DeLuca a chance to vote on the minutes that we did at the beginning of this meeting. So, we were just asking for those members who reviewed those minutes, if you voted to approve, not approve, or

abstain. So, Kyle Brothers.

DR. KYLE BROTHERS: This is Kyle Brothers. I approve.

DR. CYNTHIA POWELL: And Jane DeLuca.

DR. JANE DELUCA: I approve.

DR. CYNTHIA POWELL: Okay, thank you. All right.

PUBLIC COMMENTS

Now, we are going to move on to our public comments session, and in the announcement for this meeting, there was an open call for oral and written public comments. Dean Suhr submitted a written comment, which has been distributed to the Committee members and will be included with the minutes of this meeting. Two people submitted requests to provide oral comments today -- Rebecca Abbott from the March of Dimes is up first. Rebecca, are you on the line?

MS. REBECCA ABBOTT: I am. Can you hear me?

DR. CYNTHIA POWELL: Yes.

MS. REBECCA ABBOTT: Wonderful. Thank

you so much. Good morning, Dr. Powell and members of the Advisory Committee. Thank you for the opportunity to provide comments today. My name is Becky Abbott, and I am the Deputy Director of Federal Affairs for Public Health and March of Dimes. As I have shared in previous public comments, I had the honor of leading a group of more than a dozen public health provider and patient advocacy organizations dedicated to advancing our nation's newborn screening system through federal advocacy. Our coalition's current efforts are focused on reauthorization of the Newborn Screening Saves Lives Act, which, as we heard earlier in the meeting, expires in just six days.

During the public comment portion of the August meeting, I shared that the House of Representatives passed its version of the Newborn Screening Saves Lives Act in late July. The House bill increases authorized funding for newborn screening programs at CDC and HRSA, makes refinements to language authorizing activities at

CDC, NIH, and HRSA, and commissions the National Academy of Science Reports on the future of newborn screening. The House bill will also extend the authority for this Advisory Committee for another five years.

We are grateful to our sponsors, Representative Lucille Roybal-Allard, Mike Simpson, Catherine Clark, and Jamie Herrera Butler for their leadership on this reauthorization effort.

While the House bill moved quickly through the legislative process, the Senate has been slower to act. Senator Maggie Hassan and Cory Gardner introduced legislation in July, and since then, our coalition has been working with them and staff on the Senate Health Committee on refinements to the bill and language to address concerns from other lawmakers.

Our coalition continues to pursue all legislative options to reauthorize the Newborn Screening Saves Lives Act as soon as possible. If you have questions about the reauthorization

effort or our coalition, please feel free to reach out. I can be contacted at rabbott, A-B-B-O-T-T, at March of Dimes dot org (rabbott@marchofdimes.org.) Thank you again to Dr. Powell and members of the Committee for the opportunity to provide this update.

DR. CYNTHIA POWELL: Thank you, Ms. Abbott.

Next is Thomas Childs from the Tennessee Department of Health. We are not able to see you on the line. Are you -- are you there, Mr. Childs? If you're -- we don't hear you. So, you press star zero to talk to the operator and ask the -- ask for your line to be open.

All right. I think it's best if we move on for the sake of keeping on time. So, we'll go to the next thing, which will be bringing up the Power Point slides. We're going to move on to the RUSP Condition Nomination and Evidence Review Process. Next slide.

RUSP CONDITION NOMINATION AND EVIDENCE

REVIEW PROCESS

DR. CYNTHIA POWELL: As you may remember, the Committee has undertaken a review of our Condition Nomination and Evidence Review and Decision-Making Processes. We are focusing our review on four main areas: the nomination process, the systematic evidence-based review process, the decision matrix and decision-making process, and a possible review of current conditions on the RUSP. Next slide.

In April, the Committee discussed case definitions at the start of the review process and the need to standardize terminology regarding primary and secondary targets and incidental findings, prespecifying outcomes, and the use of intermediate outcomes such as biomarkers, range of treatment that should be included, grading the evidence, and identifying and synthesizing unpublished evidence and data.

In August, the Committee discussed systematic evidence-based review process focusing on the cost adjustments, population-level

modeling, public health system assessment, and assessing values.

Today, we will continue to focus our discussion on the systematic evidence-based review process, in particular, how the Committee assesses the impact of adding new conditions on the public health system. Next slide.

Per the Newborn Screening Saves Lives Reauthorization Act, assessing the impact of the public health system is part of the evidence review process. The assessment of state newborn screening programs is intended to evaluate the entire integrated system needed for implementation of comprehensive newborn screening, not just the ability to provide laboratory testing. The assessment includes authority, laboratory testing, interpretation, reporting, tracking, and systems for assurance of diagnostic evaluations, and evaluation of outcome. The overarching goal is to inform the Committee about the feasibility of screening, state readiness to implement new condition screening, and describe the cost of

implementing a new condition screening. Next slide.

I wanted to discuss -- sort of focus our discussion today about assessing the impact of adding new conditions on the public health system by revisiting the current decision matrix used by the Committee. There are two categories that relate to the impact on the public health system: readiness and feasibility. Although overlapping this between issues of feasibility and readiness, the Advisory Committee does not fully distinguish these concepts when evaluating capability for screening. Instead, this framework helps to assure that all aspects of implementation are considered.

I wanted to take this opportunity to review the key features of each so we can consider these features as we think about how we may refine our process. Next slide.

The key features of feasibility are the availability of valid and reliable screening tests with adequate throughput to meet the needs of

population-based deployment, the availability of systems to ensure quality implementation of the screening test that include quality reagents and data-reporting system, the availability of quality-control and proficiency-testing samples, adequate training programs for new technologies, an established approach for diagnostic confirmation available to newborn screening programs, and an established approach to long-term followup, including treatment available to newborn screening programs. Next slide.

Key features are readiness are the availability of resources for screening, diagnostic confirmation, and long-term followup including financial resources, availability of laboratory equipment, data systems, and expertise, access to specialty care and treatment, systems for data collection, and authorization for screening. Next slide.

The current approach is to assess public health impact from a population and systems perspective. Population modeling is a

quantitative approach used to compare what happens if cases are identified through newborn screening settings versus usual case identification. This approach uses data from the evidence review. Assessing cost is one component of assessing the impact on the public health system. The other is determining state readiness to implement new condition screening. The results of these three approaches are summarized as part of the evidence review process. Next slide.

Currently, two surveys are used to gather information about feasibility of screening and state readiness. An initial survey of state newborn screening programs is administered as an online survey. Newborn screening programs are encouraged to work with their partners to answer questions. A followup survey is then used to interview newborn screening programs that have a mandate to screen, have begun or have plans to begin pilot screening for the condition, or have completed a budget analysis for screening for the condition. Next slide.

I wanted to provide a summary of past Public Health System Impact Assessments that have been done. The first one for Pompe disease was a preliminary one and sort of a pilot for doing this. That was then followed by MPS1, XALD, and SMA, all included in assessment of the impact on the public health system. As you can see, the number of states that participate has increased somewhat over time, and for the last three conditions, there was fairly good participation by states. Next slide.

The Committee has encouraged the community to provide feedback on this process. We reserved time at previous meetings to gather feedback. The workgroups have discussed and provided feedback, and we've hosted a meeting of experts in the field of evidence-based review to get additional input. I wanted to recap some of the feedback we received regarding the assessment of the impact on the public health system.

Some of the feedback included that surveys may not capture the difficulties of

implementing a new condition. The overall estimates of time it would take to implement a condition -- for example, giving a range of one to three years -- could be more informative. Surveys may not account for possible impacts on primary care physicians, specialists, genetic counselors, and others. Public health programs may not know the answers for all of the questions. Others that contribute into the newborn screening system may need to be engaged. Newborn screening programs may not know at the time of the survey what a long-term followup plan for given conditions would look like, and survey questions are hypothetical and responses are subjective.

And the surveys are approved and it's a fairly detailed process to go through approval, and they are not modifiable for each condition. Next slide.

The feedback the Committee received was used to help revise the survey. Revisions were completed in 2018. The Committee has not sent a condition forward for an evidence review since the

surveys were revised, so they have not yet been utilized. The Committee received a lot of very helpful feedback. However, we don't have time today to go through all of the changes. However, I did want to provide you with a couple of examples of how the feedback we received informed revisions to the survey.

On this slide, I've highlighted some of the feedback we received from one of the Committee's workgroups. Thank you, workgroups, for all of your efforts in this.

Some of the gaps or questions the workgroup had was how accurate or valid are the answers, that more choices are needed, or do we just ask specific numbers, what are things that may inhibit you from reaching that goal, and I've condensed these into two main areas for consideration.

Estimation of the time needed for implementation activity and capturing the barriers and challenges to implementing screening. Next slide.

So, to look at how revisions to the survey can help better estimate the time it takes to implement screening for a new condition. Next slide.

So, in this instance, the question was revised and additional points in the process were added, so a more robust assessment of time needed to implement specific activities can be assessed. For example, the possible responses use to be 1 year or less, 1 to 3 years, or 3 or more years. In order to get more specific and accurate data, the response options are now 12 months or less, 13 to 24 months, 25 to 36 months, 37 to 48 months, or more than 48 months. Next slide.

Additionally, activities through the newborn screening system were further delineated to provide a more robust estimate of how much time each component of the process takes in addition to an estimate of the overall time it takes to implement a new condition, and you can see some revised list of activities were added to the survey on the right -- the updated survey. Next

slide.

So, we'll now look at how revisions to the survey can help better assess challenges faced by state newborn screening programs. Next slide.

Questions and response options were revised to improve the responses. Offering descriptions of what each category or response option means and offering open-ended questions that allow states to share more information about the factors that can impede facilitating the adoption of screening. Next slide.

So, through the review of the review processes, we have an opportunity to refine the process. Today, I'd like to discuss the current process to assess the impact on public health. I posed three overarching questions to guide the discussion.

Does the current Public Health System Impact Assessment approach of surveys and followup interviews capture all the information the Committee needs? What additional information is needed? Are there new or additional methods the

evidence review process ought to include to gather information on the public health impact? Which stakeholders are not represented in the current process? How can all of the stakeholders contribute to the information?

All right. Now, we're going to open this up for Q&A and Committee discussion. Operator, please open the lines for Committee members and organizational representatives. Committee members will discuss first followed by organizational reps. As a reminder, please use the raise hand feature in Adobe Connect, and please state your first and last name each time you ask a question or provide comments.

First, we have Jeff Brosco.

DR. JEFF BROSCO: Thank you so much, Cindy. So, I think actually the changes that we've made over the last couple of years to the survey are -- are terrific. But, it does still bump off against limitations that we really can only go as far as what people answering the survey know at the time, and Scott Shone may want to

weight into this as well. But we've done in Florida over the last couple of, I guess, conditions is we've actually asked someone to look at a lot of the questions about public health impact and actually interview, for example, subspecialists or primary care docs and try to say, well, what really would be the impact because we, as the state newborn screening program, couldn't just figure this out easily ourselves. There's a lot more information beyond that.

So, maybe one idea for us to consider is at least for three or four states, kind of dig in a little deeper and see what kind of resources are available, and maybe that might help us get a broader -- actually a deeper view of what the public health impact is and maybe Scott wants to add something.

DR. CYNTHIA POWELL: Scott, any comments?

DR. SCOTT SHONE: Sure. This is Scott Shone. I didn't want to speak until I was called on. Thanks, Jeff. So, yeah. I agree, I mean, in full disclosure, you know, I was working with --

during my time with RTI, I was working with Florida on what Jeff was talking about, and it was sort of like a life evidence review of the public health system in Florida where there was effort put into seeking out the broader stakeholders from Medicaid to the short- and long-term followup community to, you know, all the different stakeholders that you mentioned during -- during your presentation to get and share, you know, that the impact was on sharing the -- the goal was to share the information with the stakeholders and then solicit interviews to gather what their view of the impact was and then have diversity across the entire state of Florida. And I do think that the outcomes of those were pretty impactful in terms of understanding what's going to be the impact of these on their system. And so, I agree with Jeff. It goes to if you give this to one individual in the state, it's the challenge on them to then spread that, and that's -- that's incredibly hard in light of everything else that's going on. So, I think that while -- while we're

talking about the survey itself, the process, but also I'd like to throw out there again, and this is hard, you know, during the webinar, sitting next to Beth or sitting next to others and sort of talking on the sideline, what are we -- what, as a Committee, are we going to do with this? Are we actually going to use the outcomes on the condition data to inform our decision? Or is this just information gathering to share so that the programs are aware that if a condition is added despite it being, you know, despite some challenges identified in the Public Health System Impact Assessment, it's still going to be recommended, but here's what you should look out for when you go forward. So, I guess my challenge -- my additional question would be I do think -- I guess my answer to some of your questions are I do think there's a lot of information -- new information gathered. I think that if you combine this with, you know, the Readiness Tool that was presented to the Committee at a prior meeting, there are ample ways to collect the data. What's

going to drive this is if we can -- we can better -- better improve the process of collecting the data and then states need to realize that we're actually going to use it when we make a decision, or we need to decide we're going to rather.

DR. CYNTHIA POWELL: Thank you. Joan Scott.

MS. JOAN SCOTT: This is Joan from HRSA. This is sort of a followup question both to Jeff and to Scott. So, the -- the survey is an attempt to gather information where the Committee can think about broadly across the nation what the -- what it may take for states to implement. But based on the process that you were describing then, was it also a tool to help initiate conversations within the state and that it could potentially play an important implementation tool should the condition eventually get added to the -- to the RUSP?

DR. JEFF BROSCO: This is Jeff Brosco. Is it okay for me to respond?

DR. CYNTHIA POWELL: Yes, go ahead, Jeff.

DR. JEFF BROSCO: Okay. So, great question. Just a little background then. So, the way it works in Florida is when the -- when a condition is added to the RUSP, Florida has one year for its committee, Genetics and Newborn Screening Advisory Committee, to decide whether to add or not, and what we realized was that we were making that decision as a committee, and it would be better made if we had more information. And so, as Scott said, he described the kinds of things that he did, and I think you're right Joan, that just in the process of gathering that information, it started to create, you know, okay we know now we need to work with a neurologist, for example, and we, as the Title 5 agency, would say okay, we're going to have to do training for our subspecialists, because they haven't done this before or whatever those sorts of things are to actually implement the condition. It was (a) helpful to get information, so we, as a state, knew really how much resources we would need. It was also very useful, for example, for our

advocates who wanted to go to the legislature and say we want to start newborn screening for this condition, and we know it's going to require this much funds. So, it was helpful in those ways as well that it really laid out what it would take to implement.

DR. CYNTHIA POWELL: Any other questions or comments either by Committee members or organizational representatives? Kyle Brothers.

DR. KYLE BROTHERS: Yes, this is Kyle Brothers. As a relatively new member of the Committee, I just wanted to ask about the -- the opportunity to do sort of qualitative data gathering. Scott suggested sort of doing some case studies maybe with individual state programs. It sounds to me, and also from what Jeff said, that that would be really productive to do that, and I agree that some of the information that would be most useful is not necessarily a number of months, but actually having folks on the ground saying well, did you think about this, did you think about this, so I was just hoping for some

ideas or some context about whether those types of activities -- whether it's possible to build those types of activities into this process and having a, you know, we know we're going to do a focus group with the state newborn screening program or some other type of approach.

MS. JOAN SCOTT: Kyle. This is Joan Scott, HRSA. Kyle, are you thinking about something in addition to what the current process is, which is the survey to all the states? But there's also for those few states who are already implementing or have -- have pilot projects to screen for the condition under consideration, the process is to actually go in and have a more qualitative discussion with those states and not just rely on the survey because we assume to have a little more experience in states who are not currently having any activities in the area. So -- but are you thinking about something beyond that? Maybe to extend that to states who have no -- who are not doing any kind of implementation activities?

DR. KYLE BROTHERS: I don’t think -- this is Kyle again. Maybe I hadn't differentiated that far. So, I guess I don't understand the details of that part. Who -- what -- how does that conversation occur? Is that sort of through, you know, through HRSA or who -- who has that conversation? Do they come to the Committee? How does that work?

DR. CYNTHIA POWELL: Alex, do you want to respond to that? Operator, is Alex Kemper's line open?

DR. ALEX KEMPER: Alex Kemper's line is probably open, but he was sitting on mute. So, I mean, help me understand exactly what, you know, where you want me to go with that in terms of, you know, how our process works.

DR. KYLE BROTHERS: Yeah, I guess, this is Kyle, I was just wondering -- I didn't really understand that part of it, and so I was hoping to understand what you do already as a way to think about whether something else should be done.

DR. ALEX KEMPER: Yeah, uh-huh. But, I

mean, getting back to your question about sort of the qualitative part around what it takes for the newborn screening programs to get going. So, you know, we have this two-stage survey that we do in partnership with APHL. APHL really leads it. So, the first one is sort of a -- a -- goes out to newborn screening programs overall to assess their ability to do things, and then we do do a deeper dive with the newborn screening programs that have already begun to do the screenings so that we can understand some of the issues that you brought up in terms of, you know, what's, you know, what would be needed and that kind of thing.

But, my thought on this, and I think this gets to your question as well -- as well as thinking about usefulness of this process for the Advisory Committee is that there's some generalizable things that -- that are related to adopting a new newborn screening test regardless of what the particular condition is. And then there are things that are unique to each specific condition in terms of new technology and that kind

of thing or the presence of late-onset disease or diagnostic, you know, typical diagnostic confirmation and so forth. And so, one of the things that we've been talking a lot about as a group is doing some work to be able to better describe the -- the common issues and the issues that are faced each time a newborn screening test is added and then be able to focus in on the incremental stuff. So, I think I completely agree with what you said, Kyle, in terms of making sure that we describe everything that's done, and I -- or everything that would be needed to adopt a new newborn screening test. I think that the work that APHL has done to refine the survey, as Cindy talked about earlier, I think it's going to be helpful. And as we think about how we're going to use this in the future doing a better job of having sort of the commonality things described ahead of time will be useful. Does that -- does that answer your question?

DR. KYLE BROTHERS: Yeah. If I can ask one clarifying question as well. When you -- for

this deep dive that you talked about with the programs that are already implementing as a pilot project, does that deep dive involve just the laboratory people or also sort of the genetic counselors on the ground and other folks who might have a view on what the implications are for introducing that condition?

DR. ALEX KEMPER: Yeah, you know, it's -- I would love to be able to see that expanded look. We've really been focused on the kinds of things that happen within the newborn screening program and how it influences them. But I would say we have kind of a -- I'm going to use the qualitative term, and I know that I'm using it wrong, so I'm going to ask for your forgiveness ahead of time -- but this kind of, you know, snowball process where we try to identify what the big problems are and talk to relevant people, and we have these technical expert panel calls as well. But we're -- we're limited in terms of how much we can do when we do these deep dives within the newborn screening program, and some of that has to do with

time as well. But your point is well taken, and I think that we should go back into group and think about how we can expand and think about the things that happen outside of the newborn screening program.

DR. KYLE BROTHERS: That's really helpful. Thank you, Alex.

DR. CYNTHIA POWELL: Jeff Brosco, do you have a comment?

DR. JEFF BROSCO: No, Alex answered it, as usual.

DR. ALEX KEMPER: Yeah, well, you know, and I just -- I didn't mention, and K.K. just texted me about this as well to be clear, too, that as we do the survey, we make sure that we find out who gave input into it to, you know, for the written parts in terms of whether it was, you know, the person who just works within the newborn screening program versus other key members, because as they fill this out, they're supposed to have a, you know, engage a bunch of people.

DR. CYNTHIA POWELL: Yeah, this is Cindy

-- Cindy Powell from the Committee. I agree. I mean, this -- it takes time, you know, I mean I was impressed by some of the information that, you know, goes with the survey -- the new survey, that it's estimated that on average, it will take states about ten hours to complete the survey, and I think that's just maybe on the minimum side of things, so it's certainly something that is time consuming, extremely important, and something where we really want to get as much feedback as possible from as many different stakeholders as possible and, you know, I think there's a perception out there -- real or just, you know, perceived -- but that maybe, you know, not everyone has input into -- into the process. You know, we certainly are limited. The Evidence-based Review Committee, you know, is under the time constraint of nine months but just for folks to think about kind of, you know, other ways and, you know, as a clinician, they should actually focus more on the clinician's viewpoint. But certainly there are others out there -- the

public, families, things like that -- that we also have to consider.

Any other comments? I think that's it. Okay. All right. So, I think we'll finish a little bit early. Just a reminder, please check the Committee's website for updates about the Committee status. Before adjourning, I want to wish everyone a happy Newborn Screening Awareness Month. You celebrate that, hopefully, in all of your states, and unless there's any other announcements, I will officially adjourn the meeting. Thank you, everyone.

[Whereupon, the meeting was adjourned.]

[Off the record at 11:27 a.m.]