The Advisory Committee on Heritable Disorders in Newborns and Children

Virtual Meeting

10:00 a.m.

Friday, August 13, 2021

Attended Via Webinar

Committee Members

Mei Baker, MD
Professor of Pediatrics
University of Wisconsin School of Medicine and Public Health
Co-Director, Newborn Screening Laboratory
Wisconsin State Laboratory of Hygiene

Jeffrey P. Brosco, MD, PhD
Professor of Clinical Pediatrics, University of Miami
Title V CYSHCN Director, Florida Department of Health
Associate Director, Mailman Center for Child Development
Director, Population Health Ethics, UM Institute For Bioethics and Health Policy

Jane M. DeLuca, PhD, RN
Associate Professor
Clemson University School of Nursing

Kyle Brothers, MD, PhD
Endowed Chair of Pediatric Clinical and Translational Research
Associate Professor of Pediatrics University of Louisville School of Medicine

Shawn E. McCandless, MD
Professor, Department of Pediatrics
Head, Section of Genetics and Metabolism
University of Colorado Anschutz Medical Campus
Children's Hospital Colorado

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

Annamarie Saarinen
Co-founder
CEO Newborn Foundation

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

Ex-Officio Members

Agency for Healthcare Research & Quality
Kamila B. Mistry, PhD, MPH
Senior Advisor
Child Health and Quality Improvement

Centers for Disease Control & Prevention
Carla Cuthbert, PhD, Chief
Newborn Screening and Molecular Biology
Branch, Division of Laboratory Sciences
National Center for Environmental Health

Food and Drug Administration

Kellie B. Kelm, PhD

Director

Division of Chemistry and Toxicology Devices

Office of In Vitro Diagnostics and Radiological Health

Health Resources & Services Administration

Michael Warren, MD, MPH, FAAP

Associate Administrator

Maternal and Child Health Bureau

National Institute of Health

Melissa Parisi, MD, PhD

Eunice Kennedy Shriver National Institute of Child Health and Human Development

Designated Federal Official

Mia Morrison, MPH, Genetic Services Branch

Maternal and Child Health Bureau

Health Resources and Services Administration

Organizational Representatives

American College of Medical Genetics & Genomics
Maximilian Muenke, MD, FACMG
Chief Executive Officer

Association of Maternal & Child Health Programs
Jed Miller, MD
Director, Office for Genetics and People with Special Care Needs
Maryland Department of Health Maternal and Child Health Bureau

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

Association of State & Territorial Health Officials
Christopher Kus, MD, MPH
Associate Medical Director

Division of Family Health
New York State Department of Health

Association of Women's Health Obstetric and Neonatal Nurses
Shakira Henderson, PhD, DNP, MS, MPH, RNCNIC, IBCLC
Vice President, Research Officer University of North Carolina Health Board Director, Association of Women's Health, Obstetric & Neonatal Nurses

Child Neurology Society
Jennifer M. Kwon, MD, MPH, 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, MD
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, MD, MPH

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, MS, CGC

Senior Genetic Counselor Division of Medical Genetics

UPMC Children’s Hospital of Pittsburgh

Society for Inherited Metabolic Disorders

Georgianne Arnold, MD

Clinical Research Director, Division of Medical

Genetics

UPMC Children’s Hospital of Pittsburg

C O N T E N T S

P R O C E E D I N G S

WELCOME, ROLL CALL, OPENING REMARKS

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

We'll begin with the roll call. Representing the agency for Health Care, Research, and Quality, Kamila Mistry.

KAMILA MISTRY: Here.

CYNTHIA POWELL: Mei Baker.

MEI BAKER: Here.

CYNTHIA POWELL: Jeff Brosco. Kyle Brothers.

KYLE BROTHERS: Here.

CYNTHIA POWELL: Jane DeLuca.

JANE DELUCA: Here.

CYNTHIA POWELL: Representing the Centers for Disease Control and Prevention, Carla Cuthbert.

CARLA CUTHBERT: Here.

CYNTHIA POWELL: Representing the Food and Drug Administration, Kellie Kelm.

KELLIE KELM: Here.

CYNTHIA POWELL: Representing the Health Resources and Services Administration, Michael Warren.

JOAN SCOTT: This is Joan. I'm checking in for him. He'll be here any minute.

CYNTHIA POWELL: Okay, thank you. Shawn McCandless.

SHAWN MCCANDLESS: Here.

CYNTHIA POWELL: Representing the National Institutes of Health, Melissa Parisi.

MELISSA PARISI: Here.

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

ANNAMARIE SAARINEN: Here.

CYNTHIA POWELL: Scott Shone.

SCOTT SHONE: Here.

CYNTHIA POWELL: And our organizational representatives. From the American Academy of Family Physicians, Robert Ostrander.

ROBERT OSTRANDER: Here.

CYNTHIA POWELL: From the American Academy of Pediatrics, Debra Freedenberg.

DEBRA FREEDENBERG: Here.

CYNTHIA POWELL: From the American College of Medical Genetics, Max Muenke.

MAXIMILIAN MUENKE: Here.

CYNTHIA POWELL: From the American College of Obstetricians and Gynecologists, Steven Ralston. From the Association of Maternal and Child Health Programs, Jed Miller.

JED MILLER: Here.

CYNTHIA POWELL: From the Association of Public Health Laboratories, Susan Tanksley.

SUSAN TANKSLEY: Here.

CYNTHIA POWELL: And Chris Kus from the Association of State and Territorial Health Officials is not able to join us today. From the Association of Women's Health, Obstetric, and Neonatal Nurses, Shakira Henderson.

SHAKIRA HENDERSON: Here.

CYNTHIA POWELL: From the Child

Neurology Society, Jennifer Kwon.

JENNIFER KWON: Here.

CYNTHIA POWELL: From the Department of Defense, Jacob Hogue.

JACOB HOGUE: Here.

CYNTHIA POWELL: From the Genetic Alliance, Natasha Bonhomme.

NATASHA BONHOMME: Here.

CYNTHIA POWELL: From the March of Dimes, Siobhan Dolan.

SIOBHAN DOLAN: Here.

CYNTHIA POWELL: From the National Society of Genetic Counselors, Cate Walsh Vockley.

CATE WALSH VOCKLEY: Here.

CYNTHIA POWELL: And from the Society for Inherited Metabolic Disorders, Gerard Berry.

GERARD BERRY: Here.

CYNTHIA POWELL: Thank you. Today, we will begin with a panel on National Registries. This will be followed by a break from approximately 11:35 to 12:05. Returning from break, our last session of the meeting will be a

continuation of the Committee's exploration of the Newborn Screening Workforce with presentations on Laboratory and Follow-up, Audiology, Pediatric Endocrinology, and Genetic Metabolic Dieticians.

I will now turn it over to Mia Morrison, our Designated Federal Official, to provide guidance for participating on the webinar.

MIA MORRISON: 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 cannot access the audio through your computer, you may dial in to the Zoom meeting using the telephone number in the email with your Zoom link. This meeting will not have an all-attendee chat feature, but we did have a couple of comment periods scheduled for yesterday.

Committee Members and org reps, audio will come from your computer speakers, and you'll be able to speak using your computer microphone. If you can't access the audio microphone through your computer, you may dial into the meeting using

the telephone number in the E-mail with your user-specific Zoom link.

Please speak clearly and remember to state your name first to ensure proper recording for Committee transcripts and minutes. The chair will call on Committee Members and then organizations representatives. In order to better facilitate the discussions, Committee Members have been requested to use the raise hand feature when you would like to make comments or ask questions. Simply click on the participant icon and choose raise hand. Please note that the type of your device or operating system, this feature may be in a different location. To troubleshoot, please consult the webinar instruction page in your briefing book. Next slide, please.

To enable closed captioning, please select the closed captioning icon from the top of your Zoom taskbar. From that menu, select show subtitles.

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

CYNTHIA POWELL: Thank you, Mia. May I have the next slide, please.

At the February 2021 Advisory Committee meeting, the Committee invited a panel to discuss innovation and long-term follow-up. On that panel, we heard from Dr. Mary Schroth of Cure SMA, who provided an overview of the SMA Clinical Data Registry. In the past, the Committee has heard from other organizations about this topic. We heard from a representative from the CF Foundation who explained how they have sustained their CF Database over the years and funded it. we've also heard from the MBSTRN about their availability of database infrastructure.

Registries for conditions identified through newborn screening have been of great interest because of the opportunity to demonstrate the impact of early identification throughout the life course and learn how we can improve newborn screening and follow-up services for individuals who are identified at birth. We often have to rely on data from other countries to obtain long-

term data about outcomes -- countries that do a much better job of tracking their patients over the lifespan.

Developing and implementing registries is highly complex. It's also challenging in terms of funding and today, I have invited three speakers to provide us with an overview on the domestic registries used for long-term follow-up of people with hemophilia and children diagnosed with cancer.

Our first presenter this morning is Dr. Vanessa Byams. Dr. Byams is a lead health scientist in the Epidemiology and Surveillance Branch at the Division of Blood Disorders at the CDC. As team lead, she provides scientific and programmatic leadership for surveillance and health promotion activities to improve the health of persons with inherited bleeding disorders. Dr. Byams earned her undergraduate degree from Emory University and her Master's degree from Boston University, School of Public Health. She completed her doctoral training in Public Health

Leadership at the University of Illinois at Chicago. I'll now turn it over to Dr. Byams.

NATIONAL REGISTRIES FOR HEMOPHILIA AND CHILDHOOD CANCER

VANESSA BYAMS: Good morning, everyone. Thank you for that introduction. I'm pleased to be here with you today and share some information about community count. Can everybody hear me?

CYNTHIA POWELL: Yes.

VANESSA BYAMS: Next slide. Next slide, please. Thank you. Here is my disclosure.

So, first of all, I'd like to give a little bit of background, a brief overview of hemophilia. Hemophilia is an inherited bleeding disorder in which the blood does not clot properly due to deficiencies in blood clotting Factor 8, Hemophilia A, and Factor 9, Hemophilia B. About thirty to thirty-three thousand males in the United States are living with hemophilia.

Hemophilia A and B are X-linked disorders and primarily affect males. However,

women can have hemophilia too. Certain women produce very little clotting factor and can have significant bleeding.

People with hemophilia suffer from spontaneous bleeding, particularly into joints, the brain, muscles, and soft tissue.

Bleeding is also associated with injuries and surgeries. Bleeding can be debilitating across the lifespan. Repeated bleeding into joints can impair joint function and mobility and cause chronic pain and inhibit day-to-day activities and quality of life.

Preventive treatment before bleeds happen, also called prophylaxis, is critical for people with hemophilia. But the treatment itself can carry its own burden. Additional treatments for hemophilia are administered intravenously, whereby the medicine is injected in the vein, typically two to three times per week. And this can be very challenging for kids and adults in terms of long-term treatment adherence. There are newer treatments that are administered as an

injection under the skin and taken less frequently.

Although hemophilia and other bleeding disorders are relatively rare, the quality of life of tens of thousands of people in the U.S. are impacted and we have an urgent need to monitor the heath of and reduce complications affecting people living with these disorders. Next slide. And next slide, sorry, a little bit of animation.

CDC has a long history of collaborating with the bleeding disorders community to establish surveillance and monitor bleeding disorders complications.

In 1975, HRSA, Health Resources and Services Administration, received a congressional appropriation of funding to develop a program to support an integrated regional network of hemophilia treatment centers. Dr. Judith Baker will provide more details about the HTC integrated care model.

In 1983, Congress appropriated

funding for CDC to provide AIDS risk reduction services for persons with hemophilia and for others who use blood-based treatment products. During this time, CDC established partnerships with HTC, Hemophilia Treatment Centers, to develop and implement strategies to prevent AIDS in persons with hemophilia.

In 1995, CDC, in collaboration with health departments in six states, established a Hemophilia Surveillance System, which was used to conduct surveillance -- conduct active population-based surveillance to understand the prevalence of hemophilia and its associated illnesses, complications, and death. Major findings from this first iteration of surveillance was that patients receiving care at HTCs were 60 percent less likely to die and 40 percent less likely to be hospitalized for complications and were patients receiving care elsewhere.

In 1998, CDC, in collaboration with the HTCs, established the Universal Data Collection, UDC, Surveillance System to monitor

HIV and blood-born viral hepatitis in persons with hemophilia, thereby tracking blood safety and to track the prevalence of complications associated with hemophilia and other bleeding disorders.

In 2011, the cooperative agreement began for Community Counts, the latest iteration of bleeding disorder surveillance, which has an expanded focus compared to the previous two iterations.

The rest of my time will focus on describing Community Counts. Next slide, please.

This slide just shows another view of a timeline showing the evolution of bleeding disorders with the three hemophilia surveillance projects that have started at CDC. Next slide, please.

The purpose of Community Counts is to collect and share information about health indicators and complications that affect people with hemophilia and other bleeding disorders receiving care at over 140 HTCs in the US. Information is gathered about diagnoses, bleeding

events, treatments, and treatment processes, inhibitors, and more. Baseline data is collected at the initial visit and updated data is collected during annual subsequent visits.

The project also collects specimens. Up to two specimens may be collected based on risk to screen for the presence of an inhibitor and to test for HIV and hepatitis C.

Inhibitors can be a devastating complication that's related to treatment for people with hemophilia. When a person develops an inhibitor, the body stops expecting a replacement factor treatment product of the normal part of the blood and thinks the factor is a foreign substance and tries to destroy it with an inhibitor. The inhibitor keeps the treatment from working, which makes it more difficult to stop a bleeding episode. Next slide, please.

Community Counts is comprised of three main components, which I will describe briefly. Next slide.

The HTC Population Profile component

is essentially an annual head count of everyone with an eligible diagnosis that receives care at an HTC. It collects basic information on all HTC patients with bleeding disorders. This de-identified individual level data helps us to describe the overall HTC population, and you can see here a list of the data elements collected. Next slide.

The second component of Community Counts is the Registry for Bleeding Disorder Surveillance, the registry. The registry is a subset of individuals in the HTC Population Profile and it collects more detailed information, as you can see here.

Specimen collection is a part of this component and patients must provide their authorization for participation in the registry. Next slide, please.

The Mortality Reporting component collects information on causes of death. This data will be used to monitor trends in the causes of death and hopefully prevent future deaths in

the population and the data elements collected can be seen here in addition to demographic information, which is also collected as a part of this component. Next slide, please.

Community Counts is a collaborative project funded through a cooperative agreement awarded to the American Thrombosis and Hemostasis Network, ATHN, in partnership with the US HTC Network of over 140 HTCs. ATHN is a nonprofit organization whose mission is to use technology tools to advance care and research for people with bleeding and clotting disorders. Each partner has a very important role in the execution of this project. CDC provides resources, scientific and programmatic guidance, laboratory testing, and technical assistance to ATHN and the HTCs. We maintain the project data, perform analyses, and develop reports.

ATHN serves as the coordinating center for the HTCs and all surveillance project-related activities and provides the data platform to electronically record and transmit surveillance

data to CDC. It also provides training and technical assistance to HTC staff on the data platform.

Hemophilia Treatment Centers in the HS HTC Network identify and enroll patients with eligible bleeding disorders at their centers and collect patient information and the appropriate blood specimen. HTCs implement all surveillance instruments and collaborate in the analysis, presentation, and publication of surveillance results. Patients and caregivers generously contribute their information to this project. Next slide, please.

CDC and ATHN work closely with the USHTCN regional leaders through this work group infrastructure to develop, implement, and maintain the surveillance system. The executive Committee facilitates review of program goals and priorities to ensure alignment of Community Counts project activities to the requirements of the CDC cooperative agreement.

The Regional Leadership Work Group

includes regional directors and administrators, and it provides input and insight into the business, administrative, and implementation functions required for successful project execution. The regional leaders also channel and synthesize HTC and regional stakeholder input.

The Science Work Group includes some regional leaders and HTC clinicians and multi-disciplinary care providers. It provides input on clinical practice and emerging scientific issues as they relate to the Community Counts project. The group facilitates scientific review of and recommends develop of specific reports, presentations, peer-review manuscripts, and other materials to ensure dissemination of project results. Next slide, please.

When reflecting on the system strengths, I think there are several including our longstanding collaboration with the US HTC Network and partnership with ATHN. The scope and longevity of the surveillance program, including longitudinal data collection and the integration

of specimens allows CDC to track trends and important health outcomes. There is high participation of HTCs and patients and historically, the system has been flexible and able to respond to emerging health priorities. For instance, CDC expanded to add modules for children under 2, females with bleeding disorders, and quality of life. In Community Counts, these added data elements and revised data elements to capture new FDA-approved treatment products.

Our laboratory has also been very innovative in terms of inhibitor testing methodology.

As far as challenges, the treatment landscape has changed very quickly over the last few years with new treatments and gene therapy on the horizon, and these are fantastic changes for the bleeding disorders population and we're seeing a lot of great progress. But we need to make sure that we are able to keep up with and anticipate additional changes and possible unforeseen complications due to these innovations.

The Community Counts Registry Data Form Collection began on paper and it took a great deal of time and effort to develop and harmonize with the electronic data infrastructure. But we've been able to make some progress in terms of data systems modernization over the last couple of years and have made enhancements to our centralized data platform for the project.

Delays related to some of the informatics systems issues also initially hampered some of our data dissemination efforts. However, back in 2019, we were able to develop the Community Counts Data Visualization Tool, which is now available on our website. The tool displays deidentified data on patients enrolled in Community Counts in an interactive visual format.

In terms of funding, our division funding has decreased over the years, which has resulted in decreased funding to the HTCs. Understandably, the HTC's primary priority is providing excellent patient care and staff demands of integrating surveillance along with other

research and clinical studies are numerous and should be taken into account.

Most HTCs are housed within academic medical centers, so they also must be responsive to their institutional priorities. Next slide, please.

In conclusion, Community Counts is a public health monitoring program for hemophilia and other bleeding disorders. CDC's Hemophilia Surveillance Program hopefully serves as an exemplar on how to conduct surveillance for a rare complex chronic disease through a multifaceted approach. Next slide, please.

And just lastly, I'd like to acknowledge my colleagues at CDC, ATHN, and the US HTC Network who make this project a reality, and I would like to thank the participants with bleeding disorders who are enrolled and continue to contribute their data. Thank you.

CYNTHIA POWELL: Thank you very much, Dr. Byams, for your very informative presentation, and it's certainly applicable to the conditions

that we deal with with newborn screening.

We're going to hold questions until after all of the speakers have presented.

Next, I'd like to welcome Dr. Judith Baker, who will also present on hemophilia registries. Dr. Baker is Public Health Director for the Center for Inherited Blood Disorders in Orange, California. She serves as Regional Administrator for the HRSA-funded Western States Regional Hemophilia Network, Public Health Director for the 13-state Pacific Sickle Cell Regional Collaborative, also funded by HRSA, and the new Networking California for Sickle Cell Care $15 million initiative. Her expertise and research focus on how systems influence rare disorder costs and health outcomes. For the US Hemophilia Treatment Center Network, Dr. Baker co-chairs the National Patient Satisfaction Survey, now in it's third wave, and the Hemostatis and Thrombosis Dataset collected for over thirty years. She serves on the Steering Committee of the Hematology Utilization Group Study, a national

research consortium examining illness burden and costs in hemophilia and sickle cell disease.

I'll now turn it over to Dr. Baker.

JUDITH BAKER: Thank you very much for the invitation, for the panelist time, for the participation, and for that kind introduction. Next slide, please.

Here are my disclosures. These findings are my own and do not necessarily represent the official positions of the US Hemophilia Treatment Center Network, or Western States Regional Hemophilia Network, or the Center for Inherited Blood Disorders for all groups that I have had the privilege to work with for many years. Next slide, please.

So, the questions that I've posed are what have been posed to me and also to my dear colleague, Dr. Byams. What factors promote registry funding and sustainability for heritable disorders, and my answer is that regionalization is absolutely key and public health tactics need to also be considered.

As Vanessa mentioned, registries are complex interventions. They exist within individual organizations and live within a broader sociopolitical context that apply network approaches, implementation science, and the emerging science about networks is really key to the success in both hemophilia and as we're now developing in sickle cell disease. Next, please.

So, Vanessa modeled -- started to talk about the model of care, and I wanted to just briefly mention that this is a 40-year old regional model. Funding began in the late 1970s originally with 26 Centers of Excellence, but now growing within the regional structure to over 140 centers. There are a number of publications listed here about the improved survival, decreased school and work absenteeism, high school graduation rates being favorable compared to the US population, lower costs, increased employment, high quality of life and satisfaction when people with hemophilia and related bleeding disorders do attend one of the federally supported Hemophilia

Treatment Centers. Next, please.

So, the model has the patient at the center, surrounded by a core team of hematologist, nurse coordinator, increasingly nurse practitioners and physician assistants, social worker, physical therapist. But over the years, that core team has been expanded to include data managers and clinical researcher associates, which is really important to our conversation today about research and surveillance because initially, it was the nurse or the social worker who had as his or her added responsibility implementing the CDC registry and the other registries that I will talk about. But over the years, thankfully with some increased funding, that has grown to a unique position at most, if not all, of the Hemophilia Treatment Centers, and there are a variety of other consultants as needed around the table.

So, the services are diagnosis, treatment, prevention, education, counseling, outreach, our research and surveillance, pharmacy services, and care coordination. And our settings

span horizontally and vertically, the outpatient setting, the inpatient setting, and the community setting.

And what's important to recognize and remember is that these are rare disorders. Often times, in many states, the Hemophilia Treatment Center -- there might be only one serving the entire state. While yes, there are outreach clinics, often times the experts at the Hemophilia Treatment Centers, or HTCs as we call them, are often the only expert in their entire institution. So, with a small population comes the challenges of funding, of having your voice heard, of having a small voice in terms of contracting to make sure that the patients can access the Hemophilia Treatment Center. Most of them do live within large academic institutions. Some of them live within schools of medicine. Some of them are independent. Next, please.

So, why regions? Well, this is primarily a capacity issue for rare disorders. Rare genetic disorder expertise is scarce and we

share that with many, many of the other disorders that come under the purview of this Committee.

Clinical experts are isolated. The diseases are complex. And registry implementation is often a footnote.

We have found over the years that regionalization is an absolutely critical solution to building the capacity for sustainability, for sharing expertise across the geography. Regionalization, as has been implemented throughout the US Hemophilia Treatment Center Network, has this unique feature of a regional core center in each of the what you'll see is now eight HRSA regions and that the mention of a regional director and a regional administrator -- I have been serving as a regional administrator for many years -- those are key positions that are critical to building the expertise outside of the walls of the Center of Excellence. Without that obligation, there will be only patient access to the rare Centers of Excellence that occur. But with that core center and that obligation, that's

where you see the access increasing geographically.

So, the regional leadership at the core center, they are responsible for oversight, for technical assistance. We do onboarding, we provide clear expectations, we identify emerging needs, we respond to them tactically, and we build the capacity throughout our region. We do that using public health strategies, network and implementation science strategies as well. Next, please.

So, here's a look at the current iteration of the HRSA Maternal and Child Health Bureau regional structure for the National Hemophilia Program. Color coded, you see we have eight regions. Each region has a regional director and an administrator -- that was a 1990 mandate -- and these are the locations. What's interesting to notice that three of the eight regions, the regional core center is housed in a patient service organization, and that would be the Great Lakes Hemophilia Foundation, Hemophilia

Foundation of Michigan, and in the southeast, the Hemophilia of Georgia, and I think that's a great tribute to the flexibility of the National Hemophilia Program. So, we have a great diversity of patient organizations as well as the clinical centers serving as the core centers around the country. Next, please.

I'd like to nod to our regional leadership, Alisha Keehn, Kathryn McLaughlin, wonderful strong leaders who are shepherding us through all of the whirlwind changes and really very open to innovation.

The emphasis in our HRSA National Hemophilia Program Grant is access to these regional networks of coordinated comprehensive care that is provided by teams for these very complex rare disorders and underrecognized bleeding disorders. Evaluation is critical and we do that through registries as well as other evaluation at the center and at the regional level.

Quality improvement is a focus and a

priority as well as collaboration and what's critical is also that we have structured opportunities for communication across the region. Regional meetings -- there are some disciplines that get together such as the nurses, doctors, social workers, physical therapists, the data managers, Hemophilia Treatment Center administrators. That often occurs at a regional level. And also important to note, the pharmacist.

So, funding. The grant funding has been very modest on both the HRSA and the CDC side with no substantive increases and as Dr. Byams mentioned, frankly some decreases, so, only about $35,000 per HGC throughout the country of about 140 centers. That is absolutely insufficient to accomplish all of the goals and objectives of our HRSA grant or our CDC grant, which has separate funding, and it is insufficient to provide all the services that the patients need.

So, where do we get the funding? In hemophilia, over the years, that has been accessed

through the 340B Outpatient Drug Discount Program. Better than HRSA program, it is administered through the Office of Pharmacy Affairs. We are one of many federal entities that are eligible to provide outpatient drugs at these federally discounted rates. What is important is that we are one of the few of those HRSA entities that is required to reinvest the income generated back into our HRSA grant to fulfill the objectives of that HRSA grant, and there's extensive oversight in reporting back to HRSA.

So, most of the centers in our region and throughout the country, but not all, do indeed offer a 340B pricing through their outpatient pharmacies and that has been a tremendous effort. Ongoing matters regarding oversight and compliance, and we're up to the task but that has been another layer of complexity and it is some of those funds that have reinvested to support the staff that do our registry work, primarily our clinical research associates, our clinical research coordinators, as well as our data

managers. Next, please.

So, this idea that regionalization is important is not just my idea. It is thankfully and wonderfully getting increasing attention by public health experts, by implementation scientists, by academics, and I wanted to share some of that with you so that you can go back and tap into it yourselves.

This is a very nice, very brief article about *Public Health and Rare Diseases: Oxymoron No More*, and what I found exciting is that word regional was mentioned at least four times, specifically in the areas of surveillance that regionalization is really critical for rare disorder surveillance. It's also critical for developing regional centers of clinical expertise for specialty access, it's important for provider networks, for coordination across centers, across organizations. So, for example, our hemophilia population -- and I'll talk later about our sickle cell populations -- they don't only need specialty care. They need primary care too and they need

specialists outside of the hemophilia centers so that linkage is absolutely critical and best done on a regional basis.

And then knowledge sharing. Regional networks share knowledge both about their clinical care and about their databases, and that's critical for long-term monitoring.

And I want to pause and take a mention for our Regional Data Manager, Clinical Research Associate Working Group, which is something that we put together a couple of years ago where we have several people co-chair each working group and some of the needs that they identified were that they care for and oversee at least 14 other studies. So, this wonderful CDC Registry and Community Counts Program that Dr. Byams mentioned is not the only responsibility that they have. They have many other studies. These are often not full-time jobs.

The registry alone for the Community Counts Projects takes at least 1.5 hours and when you start to add in issues of diversity, equity,

inclusion, then it even adds hours. For example, if patients speak -- if their primary language is not English, that adds hours. If they are recent immigrants from another country and not familiar with Western Medicine, that adds time to introduce the concept of what is research, what is surveillance, informed consent.

So, our patients don't live in a vacuum. They often can live hours and hours away and the registries might not be their priority. Even though this year of COVID and telehealth, how we've been able to conduct the registry and Community Counts and our other obligations through telemedicine and telehealth, that's been a challenge. So, the implementing of these registries is really promoted and advanced through regionalization because on a monthly basis, our data manager and CRC Working Group gets together and they talk about these implementation challenges. They share tactics. They share what works, what doesn't work, and they're also absolutely critical for data quality -- data entry

quality. Sometimes admittedly, some of us scientists and academics can get very excited and put a lot of our effort into creating the best and most robust scientific questions in our registries but then we don't pay as much time in the implementation of the registries.

Do we have adequate staffing? How are they trained? How will we keep them involved? Often times, these positions of data managers and CRCs, they're not often long-term positions. So, how do we reduce the churn, keep the onboarding efficient, keep people engaged? Next slide, please.

So, our registry success in our region and throughout the country has been really promoted through regionalization and there are several other registries that we also do as the US Hemophilia Treatment Center Network, and I'll lightly touch on them right now. Next, please.

So, one of them is the -- what we now call the Hemostatis and Thrombosis Dataset, the HTDS, and this was originally part of our CDC HIV

Risk Reduction work, and then the regional administrators took it over as CDC moved its attention to the Community Counts project, which has just been wonderful.

So, this -- the trends here -- this is collected annually and gives us information about our demographics at our Hemophilia Treatment Centers, some of our program evaluations, and helps utilization measures, and this is just one slide about the growth in the Hemophilia Treatment Center Database over the twenty years that you see here, 1990 through 2010. And the growth has been dramatic, primarily among females with bleeding disorders. Next, please.

Another registry has been the US HTC Network Patient Satisfaction Survey. We are in the third iteration. We are very happy to say that we have reached over 5,000 patients in this third iteration and some of the colorful boxes you'll see speak to dissemination. Those are all one-page briefs in lay language to get the information back out to the patients who were so

kind to participate and to give them information directly and to further encourage them to participate in the next wave. And there's -- the box is our recent publication about the first Patient Satisfaction Survey, and this helps us fulfill our obligations to, you know, really learn about where our -- it gives us patient feedback -- direct patient feedback about satisfaction with our services, with our team members, with our care processes, and it also gives us information about barriers -- to the extent to which barriers have been a problem -- language barriers, insurance barriers, and now COVID has been added and telemedicine. That will be in the third iteration. So, this is extremely valuable information and helps us with our local quality improvement projects. Next, please.

So, I want to -- this is just one slide to show you that we actually do have a hemophilia program in Guam, which is much closer to Australia and Japan and I've had the good fortune to be part of the leadership team

developing that and keeping it sustained over the past twenty-plus years, and they do indeed participate in all of our registries despite the distance, despite the difficulties, despite their having severe health care shortages and while this might be a dramatic example, it's a testament to how regionalization is absolutely critical and can help improve access to registries so that the registries are not only available to the Centers of Excellence around the country, but people who live very far away, who might be impoverished, where insurance might be a barrier to access to the hemophilia center. Next, please.

So, I want to move in just very quickly to a couple of the implementation science and implementation frameworks that have been very useful in organizing our thinking and our work in making regionalization operational. And here, this is from Greenhalgh, you can look it up in the Millbank Quarterly. I want to ask you to turn your attention solely to the right-hand side of making it happen. That's what regionalization

does. It moves your registries, in this case for this talk, to implement registries in a scientific and orderly planned way. Regionalization is managerial, it's reengineering, it's intentional. We make the registry happen. It doesn't occur by chance. We find out where the registry is not happening, where there are gaps, where's there's staff losses -- COVID, for example, with furloughs and people losing their jobs -- and we are on it very quickly to find solutions to make sure registry work does not halt. Next, please.

This is Wagner's Framework for Creating a Regional Healthcare System, and you'll note that shared data and performance measurement is near the top, and it's absolutely critical to see that in a regional healthcare system, we're all working towards transformed healthcare, but the three pillars are critical. The middle pillar of improving healthcare delivery -- that's where most of us focus our time -- we are absolutely committed to engaging our consumers. But then there's the third pillar, which I really want to

bring everybody's attention to, which is aligning the financial and insurance mechanisms.

So, we can have a wonderful registry sitting out there, we can have patients who are engaged and believe in it, but then the insurance and the narrow networks limit our patient access to your hemophilia centers so they don't have access to engage in the registry or the Community Counts or the Patient Satisfaction Survey. That means that our registries and our data collection are going to be skewed, not as representative as we would like, and that -- we have to be very careful about that because we are dependent upon our registries to be representative so that we make sound programmatic and policy decisions. Next, please.

So, this is the collective impact model by Kania and came from the *Stanford Social Innovation Review*, that was the initial publication. This is something that was in our sickle cell grants when HRSA regionalized our Sickle Cell Treatment Demonstration Projects for

the first time in 2014, and this is -- it might look very simplistic. This is a model that was not created for health care but rather for education. But the five tactics are really what helps structure a region, and we've applied this in sickle cell and really done some really foundational work to help build a regional infrastructure there and you'll note that common progress measures are key, mutually reinforcing activities, having a backbone organization such as the regional core centers. Next, please.

And this is another model by Mary Haines. This gives some factors that should be included in models aiming to explain mechanisms of successful networks and there is an emerging causal pathway for successful clinical networks, and I would say for successful registries as well, and that's having external support, perceived leadership, internal management, and well-designed quality improvement activities, and that quality improvement extends to the registries themselves. Next, please.

And this last model that I'm going to share with you is from -- oh, next to last actually -- is the Consolidated Framework for Implementation Research by Damschroder and it's really important to take a look at all of the areas where a registry can be viewed as a complex intervention and that attention needs to be paid so that it is successful. Next.

And this is the last slide for the different types of implementation frameworks, and this is by Rycroft-Malone and noting that facilitation and resources are absolutely critical and how that manifests to successful networks. Next.

I'm going to roughly go through some of our sickle cell slides because I know that sickle cell is one of the heritable disorders in this group's purview. We did indeed have the honor of being awarded the HRSA Sickle Cell Treatment Demonstration Project in our Pacific Region. There are five regions in the country for sickle cell, and we have thirteen states, and

those are our clinical leads as well as our community-based organization leads. Next, please.

One of the key matters is how can you -- where are you going to have your registry located. If there are insufficient members of clinical centers, then you're going to have again a very truncated view of the population you wish to study. One of the first things we did was to build a new sickle cell center in South Los Angeles, where there had been none, and that is what you see here. Next.

Why Los Angeles? Because over half the California adults with sickle cell live in LA and the mortality had been higher and there were no centers. Next, please.

From that, we -- our HRSA grant we were re-awarded, and one of the requirements was creating State Action Plans, and most of the state in our region did, and we created our State Action Plan for California in 2018 and one of the key priorities was surveillance and registries, not just clinical care. Next.

From the State Action Plan, as a background document, it is a patient voice and clinician voice, an advocacy voice, we are able to successfully go to our state legislature and educate our policy makers and successfully obtain $15 million as part of the governor's budget in late 2019, and that's what created Networking California for Sickle Cell Care. Next.

And this three-year project -- we're finishing year two right now -- has four prongs on it and we are working in those areas simultaneously. So, it's not just building a clinical network, and we are now very proud that of the five adult centers that we promised to create, we are now nearly up to ten clinics.

Our focus is also on workforce. Our focus is also on surveillance as well as outreach and education. Next, please.

And this is a copy of our first-year report, and in the third box down, you'll see our surveillance and data collection efforts, and they include not just registries, but also a data think

tank to bring together a diverse set of stakeholders who would never get together previously. There was no structure, and we provide that structure as well as real-world qualitative evidence and case management system that's unified, another registry for our sickle cell disease community-based organization and on the CHWs, the community health workers. Next, please.

And as I wrap up, I wanted to briefly show a version of the CDC's ten essential public health services and, of course, registries and evaluation, that is part, but our success over the years has been access to a registry needs to be embedded within the entire public health framework for rare disorders. And one of the areas missing, but has been updated since this slide, has been diversity, equity, and inclusion in the public health services. That is absolutely critical for successful registries implementation as well as long-term sustainability. Next, please.

So, as I wrap up here, I want to go

back to where I started, which is that investing in a regional approach for capacity and sustainability is key. The funding for registries needs a regional approach. It should not be up to the individual centers where the patients go where they might be enrolled in the registry. Those individual centers for rare complex disorders alone do not have sufficient capacity, bandwidth, expertise, clout to obtain and sustain sufficient funds for registry maintenance, invest in a regional approach. It's a very practical innovation. We've got a lot of successes and proof of principle. Next, please.

And this is a bib of all of the articles that you might find interesting on the matters of regionalization.

And next, I want to thank you very much for your time. I'm available to answer questions at the end. Thank you.

CYNTHIA POWELL: Thank you very much, Dr. Baker. You really got down to the crux of the issues that we're dealing with. As I said, we'll

hold questions until after our next speaker.

Our last panelist is Dr. Lynne Penberthy, who will present on the National Childhood Cancer Registry. Dr. Penberthy is the Associate Director for the Surveillance Research Program, which is within the Division of Cancer Control and Population Sciences at the National Cancer Institute.

Prior to her NCI appointment, Dr. Penberthy was the Director of Cancer Research Informatics and Services and Associate Professor of General Internal Medicine at the Virginia Commonwealth University Massey Cancer Center. Dr. Penberthy was also involved in biobanking and annotation of specimens using clinical data. She has twenty years of experience in cancer surveillance and automation using secondary data.

I'll now turn it over to Dr. Penberthy.

LYNNE PENBERTHY: Good morning. I just want to do a sound check. Are you able to hear me?

CYNTHIA POWELL: Yes.

LYNNE PENBERTHY: Okay, great. Thanks. We've had some technical difficulties. So, I am not going to be sharing my video, for which I apologize. But I don't think that's essential.

So, thanks so much for giving me the opportunity to talk today. I'll really be speaking primarily about our National Childhood Cancer Registry or NCCR and some of the challenges and solutions that we face in developing this infrastructure.

So, the objectives that I'd like to accomplish are really to briefly describe the National Childhood Cancer Registry, it's purpose and goal, to illustrate some examples of specific challenges that we have faced related to initiating the Childhood Cancer Registry, particularly because these are rare diseases and focusing largely on data access and privacy issues. And then lastly, I'd like to describe some methods and considerations that we've used as

the NCCR is being developed. And this is very much a work in progress just to let you know.

So, the purpose of the NCCR is to leverage and link disparate data sources from different data sources to create an infrastructure that can help us better support research on childhood cancer.

The core data for the system are derived from cancer registries, but we've expanded those data to include additional very relevant clinical information such as detailed treatment, genomic characterization of the tumors, and to try to capture the trajectory of care for each cancer patient from diagnosis throughout their life and especially for kids, this includes capturing multiple primary cancers and recurrence of their disease. We're also linking in other relevant factors related to risk and outcomes such as residential history and social determinants of health. Importantly, we're integrating this within the Childhood Cancer Data Initiative or CCDI, which is a new initiative that's sponsored

by the National Cancer Institute and represents a federated data ecosystem that is under development.

So, the registry, as I mentioned, leverages existing data sources that allow us to capture information on all pediatric and young adult cancers in the US. It's important to note that we're accumulating these data through linkages with cancer registries, and cancer registries are somewhat unique in that they are population-based. That is, they capture all cancers within a defined geographic area, at least in theory, and the registries maintain patient identifying information and have the ability to incorporate data on all childhood cancer cases longitudinally.

Reporting to the registry is HIPAA exempt and all health care providers are required to report information on cancer as the diagnosis, treatment, and outcomes to that state or general regional registry. And, in fact, every state has a regulation that requires this. They differ

slightly from state to state, which is a bit of a challenge, but each state has that regulation.

This is just a summary of the current participating registries, which represent about 77 percent of all US childhood cancer cases from twenty-three states. We're trying to expand that over time.

And the next thing I'd like to talk about a bit are some of the registry components that are really critical. As I mentioned, we have routine linkages, which will be performed centrally via an Honest Broker with external data sources, and the first of these are to capture complete abstracts on each cancer case plus text documentation from 1995 through the current date. The text documentation is important because that permits us to use natural language processing or artificial intelligence to structurally extract key treatment information, and this is a work in progress. We're not finished with this as of yet.

We also link with the National Death Index, State Vital Records, and importantly, we're

currently linking with Lexis Nexis and again, this is performed centrally, not by each state, to capture residential history routinely and biannually. This is really important for these kids because they survive fortunately for many, many years often, and it allows us to perform linkages on a longitudinal basis as the patient's address changes over time.

We're also looking to capture Financial Toxicity. We have not done this yet. But this will provide data to help us understand the impact of cancer on patients and their families.

The other critical linkage that we're performing is what we call the Virtual Pooled Registry, and this is an infrastructure that's been developed that supports linkage across all cancer registries that will enable us to capture subsequent cancers to annual linkages with all the registries in the US.

The Planned Central Linkages that I'd like to mention are pharmacy data with CVS,

Walgreens, and Riteaid, which we have in near real time for the SEER Program. We also have data from United HealthCare, which includes the Pharmacy Benefit Management System from UHC.

We're beginning to capture longitudinal detailed radiation oncology data, and this is important not just for the initial course of therapy, but also to capture information on treatment of recurrences.

We have claims data linkages that allow us to capture that detailed treatment and comorbidity with United HealthCare. The linkage is right now in process even as we speak and we're proposing to link with a large subset of the Medicaid data later this year.

We've been working to capture radiology reports and images to use for both case finding as well as again identification of recurrent disease. And we're working with several partners on this including Ambra Health, AIM, which is a subsidiary of Inspirada, which we've been using for e-path reporting, and also we're

working with selected state cancer centers who are providing us data.

As I mentioned, we're capturing genomic data and we're currently in discussions with Foundation Medicine and Caris Life Sciences and we do collect some individual biomarkers that are available from the pathology reports.

The other important data linkage that I'm sure that you all resonate with is birth records, and that's very important to allow us to capture things like parental address at birth, which will allow us to have a more accurate residential history for these patients prior to their age 21 and also to allow us to identify critical issues that may have happened at birth such as the Apgar score, whether or not the mother smoked, et cetera.

This is just an example, and I wanted to show you to sort of give you a sense of the magnitude of the potential value of these data. This is SEER-linked pharmacy data from 2013 to 2020, and this is really for adults and pediatric

patients, so I didn't break this out. But I think that it really illustrates the point here is that this -- this first box on the left is tyrosine kinase inhibitors or TKIs and we have more than 188,000 fills for more than 20,000 patients in our system.

For PARP inhibitors, which are very important for breast and ovarian in particular, we have three agents that we've captured on over 1,000 patients with 7,000 fills and, of course, the CDK 4/6 inhibitors, we have one of those, and we've had more than 40,000 fills on over 4,000 patients. So, I think this gives you a sense of the magnitude, and we'll be doing this similarly for the pediatric cancer cases exclusively.

Next, I'd like to move on a little bit to the infrastructure. So, looking at the workflow process and the necessary data platform consideration for a system such as the NCCR.

So, this is really a complicated slide, and I'm not going to spend much time on it, and I'll share these slides later. But really,

it's just the conceptual framework for the NCCR and I think what's important here that I'd like to point out is that we have data sources that are just routinely and currently linked on the left and some of the more innovative or pilot data sources on the right. And each registry that is participating in the NCR -- NCCR has it's own virtual server within an enclave hosted by our contractor, Information Management Services.

And so, their data will be maintained in their individual servers including PII that will permit these linkages and then the central component of the NCCR will only have de-identified data and those are the data that will be acceptable potentially to researchers and individuals.

This is just sort of another way of looking at that to really look at the flow for the data into the central repository and again to highlight, each of the registries has their own individual virtual server including the PII and that will allow them to have these additional

linkages and then they will submit their de-identified data to the central repository.

I wanted to take a moment to talk about the NCCR data platform. This is incredibly important to us at NCI because it's somewhat specialized and -- and it will require that the data products that we develop and the data access process can be overlain on the system. And so, the platform actually needs to support cohort discovery, simple linkages, it needs to protect privacy, and also to provide a governance structure for allowing people to access various components of the data and data products.

We had an RFI that went out about, I think, eight months ago and we got eighteen responses to that, which was very good, to allow us to identify potential applicants and generate ideas for the RFP, which we're in the process of developing and should be out in the next three months.

I'd like to move on really from that to talk about some of the data access and release

process considerations that we have for our data.

So, we have some underlying goals that -- for the NCCR -- that are very similar and align with what we do for SEER and -- and really, our policies and processes for sharing cancer surveillance data with researchers across all registries within the SEER program. And it's part of our mission at NCI to allow access to those data, but it has to fit within the NIH data-sharing policy framework as well.

Of course, we need to protect patient confidentiality and privacy and reduce the risk of re-identifiability and I'll talk more about this in a moment.

And then one of the things that we struggle with a great deal is minimizing the risks for inappropriate use of the data including analytically inappropriate use because we don't want people coming to incorrect conclusions because they're not using the data correctly.

And so, there is some special consideration for the NCCR that we had to think

about. The first is because these are kids, they have a high risk of re-identifiability because they're rare tumors. As we increase the breadth and depth of data that we have on each patient, that increases that risk of re-identifiability.

Another important consideration is that, as you all know, in recent years, individual computational capability is just expanded dramatically, and so, this provides an opportunity for mal-intended persons to develop algorithms for re-identifying patients and in particular using such things as open websites that might permit possible linkages such as GoFundMe sites to enable re-identification. Even though the dataset that we make available is de-identified, there's an opportunity for these rare tumors to re-identify individuals.

And so, some of the solutions that we have in place or are developing include a tiered system for data release that has the potential to require IRB review. We now have a central IRB contracted for the SEER and NCCR program.

We also are developing and have partially developed a data release system that's linked to the central authentication and authorization process at NIH, the eraCommons system.

We also have hired and have a contact with an external consultant, who is helping us to formally assess the risk of re-identifiability and advising us on the steps that we need to take for risk mitigation, and I would suggest that this is a very important thing to think about for any childhood registry.

And then lastly, the other thing that we've heard from all of our confidentiality and privacy experts is that regardless of the -- the steps that you have in place to protect privacy, you still need to have people sign a data use agreement that says that they will not try to identify anyone.

So, our goals at the NCCR are really internal and external. The internal goals are allowing us to share data between the central

registries who submit the data for de-duplication purposes, completeness of reporting, as well as identification of multiple primary cancers, to look at quality control, and then to provide the central registry access and use of the linked data.

The external goals are also very important and really to help us understand the applicability of the Common Rule and Public Health Reporting for Surveillance and to enable multiple models of data access that will allow us to maximize patient privacy but promote data utilization and research, and as I mentioned, we have a user authentication and authorization system and we have a number of restrictions in tiers for access and release of the linked data.

And then lastly, this is the most challenging to some extent, and that is developing a criteria to evaluate fitness for use of the linked data. That is, what are the types of research that the data would support.

And so, in order to meet these goals,

as I mentioned, we have developed in part a new multi-tiered authentication and authorization process, which has four tiers.

The first tier for the FIT program is really a completely de-identified data that does not have any dates or geographic variables including registries, and this system is available with minimal data use agreement, and it's currently live. So, this can be downloaded by anyone across the world.

Tier 2 is a limited dataset. So, it has some dates with minimal detailed characterization variables, and this is also live and has a slightly more robust authentication and authorization set of requirements.

Tier 3 is again a limited dataset, but this includes some special variables that we don't routinely release as part of Tier 1 or Tier 2 including biomarkers, multi-gene panels, et cetera and this is live. And what we've done here is to ask investigators to submit a brief proposal on what they're proposing to use the data for and

that's reviewed internally to assure that the questions that they're asking can be supported by the data.

Tier 4 is currently under development, and that is again a limited dataset, but it has the longitudinal treatment information for each patient. So, again, that increases the risk of the identifiability and in some cases may require IRB review.

And just to reiterate, each tier has a requirement to sign a data use agreement that's targeted to the level of data that are released.

I wanted to mention that one of the things that we are also doing because we want to share the data is developing data products. And again, we have an incremental and tiered system that we're developing with five levels of data. That includes a really minimal level that sort of counts and indexing that people can use, for example, if they were putting in a grant proposal or they just wanted to know how many cases there were.

And then the second tier is what we call ready statistics, and these are largely interactive tools and we're developing a system called PEDS Explorer, and if you're interested, you can see what this is going to contain at this link listed here.

The third tier is really what we call canned analysis, and that's using her SEER Stat program, which has increased flexibility, but it still has some limitations in terms of the types of analysis that can be done on the data.

The fourth tier, which we are developing, is a cloud-based system of analysis using things like SPSS, SAS, et cetera. In that case, the data would not be downloadable, but the person -- the individual researcher could do the analysis online.

And then the fifth tier, which is for downloading data, is likely to require IRB approval. And what we found is that in some instances, people actually have to download the data because they need to be able to do things

that might not be supported in the cloud.

This is just an example from the PEDS Explorer. This is our current static report, part of the initial data product, and really it looks at the incidence rate of cancer -- specific cancers denoted by the various colored lines at each individual age group. And what you can see is that the incidence for many of these increases in the later adolescent years. But I think this is just an example and I'm happy to share the weblink for this report if you're interested.

I was asked to talk about a few other considerations that we're trying to address that might be relevant for other types of registries. And one of these, I think, is really related to the prior presentations, and that is special considerations because of health department reporting.

As I mentioned, you know, there are regulations in each state, but there is some variation from state to state, and what we found is that it's really essential for registries to be

directly engaged with the state legislature and with the Department of Health. That -- for us, the registries are often located either at the Department of Health or at a university, often that has an NCI-designated cancer center.

And, in fact, what we found is that it's very common practice for our registries to work directly with the state legislature annually to create or modify reporting requirements because, as you know, the reporting requirements evolve over time, and so that relationship has been very helpful in order for us to be successful to be allowed to capture the data that we need for surveillance.

The last thing that I wanted to mention, which this is a very complex topic, and I can only just barely touch the surface here, and that is privacy preserving patient linkages.

Physically, these use patient-identifying information and they hash and tokenize the PII to be able to permit encrypted linkages without releasing PII. There are tons of

companies who have these capabilities, and in fact, we at NCI have performed recently a landscape analysis and in that landscape, twenty-seven companies are reviewed and the report is available if you're -- if you're interested. From that review, four companies kind of floated to the top as something that we were interested in pursuing further, and these are currently under evaluation using a formal assessment process for accuracy, sensitivity, and specificity, and it's based against a set of gold standard manually validated and linked datasets.

So, in summary, really the takeaway from that is that, you know, if you can, link PII because it's always optimal. The hash tokenization is not a magic solution. I mean, it does work well, but you have to be very careful about that because linkage results vary depending on the completeness, the quality, and the type of PII that are available in each of the datasets that are being linked. I mean, I know that seems obvious, but it's very, very important.

I also wanted to mention that often, the companies are willing to work with you because we found that some slight customization and variation of the methods that they're using can improve the accuracy of linkage using these P3RL products.

And with that, I will end. Thank you so much for your attention.

CYNTHIA POWELL: Thanks very much, Dr. Penberthy.

I will now open it to questions from Committee Members first, followed by organizational representatives. And again, as a reminder, please use the raise hand feature in Zoom when you would like to make comments or ask questions. And when speaking, please remember to unmute yourself and state your first and last name each time you ask a question or provide comments to ensure proper recording.

As we give people a chance to get their hands raised, I was wondering, I'd like to ask Dr. Baker are you able to give a little more

detail regarding that 340B pricing that's utilized to help fund the hemophilia registries?

JUDITH BAKER: Sure. 340B pricing -- it's the 340B Drug Discount Program and it's also called PHS Pricing or the VA Pricing Program. It was a law passed in 1992 to provide eligible federal agents -- entities an opportunity to purchase outpatient drugs at federally discounted prices. It is again a HRSA program. It is administered within HRSA's Office of Pharmacy Affairs. The Hemophilia Treatment Centers who receive a -- who are part of HRSA's National Hemophilia Program are one of those eligible entities. We are one among many. Other eligible entities include Federal Family Planning Programs, Black Lung Clinics, a proportionate share of hospitals. It's a very -- it's a growing and complex program. The prices can only be used for the purchase of outpatient drugs. Only the 140+ centers that are contracted with the eight HRSA hemophilia core centers and the core centers themselves are considered eligible entities. So,

I'll just stop there as a very brief overview. Much more detailed information is on the OPA, the Office of Pharmacy Affair's website.

CYNTHIA POWELL: Okay, thank you.

JUDITH BAKER: My pleasure.

CYNTHIA POWELL: Shawn McCandless.

SHAWN MCCANDLESS: Thank you. Shawn McCandless, Committee Member. There's a long list of question. Cindy, thank you for your question. That was one of mine that I may come back to for more information.

I guess the first question for both of the first two speakers is maybe what -- what -- what would be the cost of setting up a similar system for other kinds of newborn screening disorders that -- from the beginning and sort of a related question is did the HTC and the ATHN predate the availability of the federal funding? And then sort of a third question, if anybody is willing to take a stab at it is, is it valuable -- is it necessary to reinvent the wheel every time a different sort of group wants to set up a registry

or is the infrastructure that's already been built robust enough or flexible enough to expand to incorporate other types of disorders?

JUDITH BAKER: How come you ask the easy questions? Vanessa?

VANESSA BYAMS: Yeah. Yeah, I was going to try to take a stab. So, going, I think, to the second question about did the US HTCN and ATHN predate the federal funding for bleeding disorder surveillance. So, yes. The HTCs have been in existence since 1975, and the CDC funding got surveillance came later. And actually, the partnership with ATHN began in 2011 at the start of Community Counts. So, prior to that, CDC funded the regions directly on behalf of the HTCs and with the current cooperative agreement, CDC funds ATHN, who funds the region, who funds the HTCs and others mentioned in my presentation. ATHN, as an organization, is responsible for not only the coordination of the program at the HTCs but also for data-captured infrastructure that we use for the electronic data collection.

For question 3, I think related to the infrastructure. I do feel that we have a solid infrastructure that could be adapted, you know, other than hemophilia, you know, we collect data for more rare bleeding disorders, other rare clotting factor deficiencies, Von Willebrand disease is also a bleeding disorder that we cover. I know in years past, we've talked about, you know, whether or not to think about the HTC as a sort of hub not only for bleeding disorders and hemophilia care but also for other blood disorders, and many of the HTCs do provide care for venous thromboembolism, clotting, sickle cell disease, as Judith talked about in her presentation. So, I think it depends on the center and the region in terms of what the patients that those centers care for. But I think the model and the infrastructure is there and is strong and historically has been strong.

As for the overall cost, I think because it's been such a longstanding program, that's a little bit hard to estimate and I think,

especially in recent years, as far as trying to not only fund the HTCs, but also as we try to enhance the technology and the technological infrastructure and as I mentioned we began on paper. So, we were, you know, kind of coming from a more antiquated model and trying to -- to, you know, modernize appropriately. I mean, I would say, you know, gosh, tens of millions. I don't know. I may be sort of overspeaking. But that's my estimate. Right now, we fund ATHN for Community Counts $4.3 million per year, and so that -- part of that, you know, stays with ATHN, part of that goes to the HTCs and Judith did a very nice talking about what that covers and what that doesn't cover. And I think, you know, certainly at the regional level, there's been a lot of investment over the years and they have worked to have HTC staff and patients and families invested and interested in contributing, you know, long term, which we are very thankful for. So, I think a lot of our success, you know, has been despite -- despite the amount of funding that we

have provided over the years.

Judith, did you have anything to add?

JUDITH BAKER: Yeah. Thank you, Dr. Byams. You really hit it. Just a few things, and thank you for the questions, Dr. McCandless.

On question number 2, in terms of the timeline, actually ATHN was crated in 2006 by a diverse group of stakeholders including the Hemophilia Treatment Centers, industry, government partners, patient organizations because we saw the need for a national secure database infrastructure as we were moving away from paper and pen. So, we -- the collective we -- created ATHN, and we are extremely proud of that. And the CDC, yes, used to fund the regions directly starting back from 1990 to do HIV risk reduction in the HIV tragedy in hemophilia. And at a certain point around 2012 or so, CDC, you know, we talked and there was a move, my understanding is, to reduce administrative costs, and the CDC let us know that and we asked ATHN, the organization that we created, to help us write and submit one national

grant on behalf of all the HTC regions to participate in the CDC new Community Counts Surveillance, and we were very fortunate to have been awarded over these years. So, it's always been a wonderful collaboration.

And the other thing in terms of reinventing the wheel, that's why I brought in sickle cell to this particular talk. It's a real thin diagram in terms of the number -- the hematologists who are trained to take care of blood disorders, they have training in sickle cell and hemophilia and a wide variety of inherited clotting disorders as well, and at more of the smaller centers and mid-sized centers is where we see the same exact team that we sometimes call the Hemophilia Treatment Center but they're also caring for sickle cell disease.

At some of the larger centers -- well, the mid-sized centers, you might see the same physician who cares for both sickle cell and hemophilia, but they may have a separate nurse, a separate social worker, a separate ancillary set

of core providers. At the larger centers around the country is where we see completely separate sickle cell and hemophilia centers. And we've done some very soft qualitative data collection whenever we go around asking to other regions -- other hemophilia center regions for presentations, we ask how many of you take care of sickle cell, and very soft data, we could do a better job of it. But at least a third -- a fourth to a third of the hands raised. So, there's great overlap and it's because of that that we think you do not have to reinvent the wheel. We do have structures in place. Regionalization works, particularly the rare disorders. The phrase that I've been coining is mobilizing across blood disorders because there's such similarities in terms of the patient complexity, the workforce -- the clinical workforce who knows how to take care of this, the team-based care that's required, the policy issues in terms of being -- having a rare disorder and you are -- the patients are not guaranteed access to the specialty centers because they are rare,

they have very little power to contract with managed care organizations, both public and private. So, at the policy level, the systems level, the patient care level, at the provider level, and often at the CBO level -- the community-based organization level -- there's great similarities between many of the rare disorders, in particular the blood disorders.

But the cost, I will leave that to Dr. Byams' estimate. Thank you.

CYNTHIA POWELL: Dr. Penberthy, did you want to respond?

LYNNE PENBERTHY: Oh, I just wanted to add a comment to that. One of the things that we have done in terms of collecting data and looking at patients who are treated, perhaps, outside Centers of Excellence, is to link to existing claims data, because there's a tremendous amount of information available in the claims data. In particular, we have a project that we've been trying to move forward working with the National Heart, Lung, and Blood Institute where

they have identified from Medicare and Medicaid, all patients with sickle cell disease, and they have longitudinal history. As you know, many of those patients are in Medicare or Medicaid because of disability and it allows to track those patients for the long-term and really be able to look at who is treating those patients outside of those Centers of Excellence. So, just something to think about as you're developing these registries to try to leverage existing data sources, particularly federal data sources that might be helpful.

CYNTHIA POWELL: Thank you. And in order to link to that data, do you have to utilize or do you utilize ICD-10 codes to do that?

LYNNE PENBERTHY: So, basically, what we do is link at the patient level. So, you have to have patient identifiers in order to link. Medicare and Medicaid, we've been linking SEER Medicare for many, many, many years now, and this is something that NHLBI did and the reason that I got involved is because, as you may know, there is

some question about the risk of hematologic malignancies in patients -- sickle cell patients being treated with some of the new therapies. And so, we're going to be linking our data, hopefully, with that -- that cohort of sickle cell patients from Medicare and Medicaid to look at the risks -- the overall risks of other hematologic malignancies in those patients who have, you know, rapid bone marrow turnover and may increase their risk.

But my point is that you do need to have PII. So, that's something that if you don't collect that in your data system, it wouldn't work.

But, you know, we use -- so there are ICD-10s, which are good for diagnosis, but also what's really important are the CPT and HCPCS codes, which are very specific to, you know, generic drugs, other types of treatment, and they're really quite accurate because those are things that physicians and providers get paid based on, and so there is some overview to make

sure that they are, in fact, correct.

CYNTHIA POWELL: Um-hum, yeah. I asked that because there's a lack of ICD-10 codes for many of the rare disorders that we deal with.

From the Committee, Mei Baker.

MEI BAKER: Sorry, I had to unmute myself. Mei Baker, Committee Member. My question is for Dr. Penberthy. I just wanted to ask, in your registry, if it ever has been discussed about biobank?

LYNNE PENBERTHY: Funny you should ask. Actually, for the larger SEER program, which includes adults patients, we have what we call the Virtual SEER-linked Bio-Repository Project. This is a very large pilot study that we're doing to allow the registries to sort of have a virtual biobank. And what that means is that, you know, we know about all the cancer cases, and so, our registries have very good relationships with the pathology labs that submit the information to the registries, and so, they can identify potential specimens. Largely, these are formalin 6 parafin-

embedded, so, you know, that's just within that purview.

The other thing that some of the registries are doing, which is similar, is what we call a Residual Tissue Repository, and for those of you who are not familiar with pathologists, the College of American Pathology requires pathologists to maintain the tissue blocks and the slides for ten years. After that ten years is done, they can throw them away. And so, some of our registries are actually working with the path labs to collect and hold those residual or discard specimens. So, yes, indeed, we are looking that that and are hoping to expand that beyond the pilot phase.

MEI BAKER: Thank you.

LYNNE PENBERTHY: Did that --

MEI BAKER: Yes.

CYNTHIA POWELL: And we'll open it up to organization representatives. Natasha Bonhomme.

NATASHA BONHOMME: Great. Natasha

Bonhomme, Genetic Alliance. These are great presentations and I really feel like I've learned a lot and I really appreciated the discussion about data products and just the sophistication around that. That's really great that you have that and have it laid out in such a way that if you need data, you know which category you fit into. So, really kudos to that.

My question is for Dr. Baker and is also about 340B, something that a year ago I knew nothing about and now that I'm on the Board of an FQHC with a pharmacy, I know more than I ever thought I would know about it, you know, and from those discussions, knowing about the complexity of the program and, I think, what someone called controversy or just the ups and downs of it and knowing, you know, some feeling the need to talk about potentially other revenue sources or, you know, what to do because it does really contribute so much, and I just kind of wanted your take from the work that you're doing, kind of where those conversations are. Are you thinking about the

same things or is it kind of it feels more settled in the work that you're doing?

JUDITH BAKER: Thank you for that good question. Not settled. Repeat, not settled. It's an ongoing effort that takes a great deal of thought, coordination, expertise. We're fighting it. You know, it's -- it's sustainability. It's Hemophilia Treatment Center sustainability. It's rare disorder sustainability. Nobody can do this alone. Partnering is absolutely essential. Being very strategic and having frankly sufficient capacity to address these policy matters is -- is absolutely critical. We have, you know, all of the hemophilia regions have a different number of states and it's a real stretch to monitor all of the -- let alone the state matters -- state Medical policy changes -- Medicaid is critical. Over half in California -- I'll just speak about California. Over half of our hemophilia center patient population is insured by either Medicaid or two other state and sometimes state-level

federal programs that follow Medicaid reimbursement policy. That's over half our population. So, we absolutely must be proactive in monitoring any potential changes to pharmacy reimbursement rates that could unfortunately negatively impact the revenue that's available to the centers.

And then there's private insurance that can create narrow networks as well as their own pharmacy benefit managers who completely cut out hemophilia centers.

So, this is an extensive, very complex matter, which requires great collaboration with the team members, not just within hemophilia at all, both in broader policy fields, but also to really identify what is the true cost of the care that's provided. So, we've had to cost that out, and we've had some success in having the state of California recommend a revised reimbursement rate that as new products advanced, such as the bio similars, which are provided by injection, there's great diversity of how that is reimbursed.

And the lack of coordination, lack of optimal reimbursement -- I won't say adequate -- I will say optimal -- I mean, the bottom line is there must be optimal reimbursement for team-based care for people with complex disorders, particularly if those are rare and inherited. So, this is an ongoing matter. 340B has been great. It's wonderful. It's still there, but this requires ongoing significant attention. It's the financing of outpatient care for our rare disorder populations.

CYNTHIA POWELL: Thank you.

Debra Freedenberg.

DEBRA FREEDENBERG: I just had a quick question for Dr. Penberthy. It's related to the biobanks. When you're linking to the biobanks, does that then become a consented registry or is that still not considered a consented registry?

LYNNE PENBERTHY: That is a great question and I'm very sorry you asked. No, I'm kidding. Seriously, we have -- we have developed

this pilot and the way that it is working is that the registry, you know, the folks at the registry serve as an honest broker. So, they link the clinical data with the specimen information in a de-identified way. And so, the investigator who is doing the tissue analysis never gets PII. So, because the registry is an honest broker, they're not involved in the research, they've been able to do this.

And the reason I say it's a challenging question is because we're proposing to move this forward; however, the NIH has a genomic data-sharing policy that is somewhat in conflict with the common rule in that we want all people to share genomic data who have been funded by NIH into the GDC, the General Data Commons. However, they are requiring consent. And so, we're -- we're trying to really understand, you know, is there a way that we -- we can get an exemption for this, and in part because the whole idea of biobanking is incredibly important and most of the tissue research, at least for cancer, is based on

clinical trial. It's a really biased subset of patients, right? They're younger, they're, you know, they have no comorbidities, it's not the real world. And so, really, the value of this is that you could look at specialized sub-population, so, you know, minorities with this population-based system for accessing tissue. And I think that that's really, really important because we don't know whether there are differences in some of the results that are seen based on clinical trials and, you know, minorities don't enroll in clinical trials. We've been trying to address that for many, many years.

So, the consenting question is a very real question and for our purposes at the VBR, it's not really possible because the reporting to the registry, as you know, is required by law, and there's no -- there's no informed consent for that because, you know, it's HIPAA-exempt and it's required by law. But so, there are ways, I think, to do this, and we're still working through some of those issues. But clearly prior to 2015, it's

not a problem from the NIH perspective as well, and those are the studies that we've been doing. So, does that answer your question?

DEBRA FREEDENBERG: Yeah, thank you.

CYNTHIA POWELL: Jed Miller. Oh, go ahead. Did you have something else, Dr. Penberthy?

LYNNE PENBERTHY: Yeah. I was just going to say that if -- if anybody is interested, you know, I'm happy to -- to share information on the Virtual Bio-Repository and where we are and what's been done and so on. I've got some slide decks that I'd be happy to share.

CYNTHIA POWELL: That would be great. Thank you. Jed Miller.

JED MILLER: Yes, hi. Jed Miller from Association of Maternal and Child Health Programs. I'm wondering if any of the panelists have any comments on state health information exchanges. I'm curious, Dr. Penberthy, if they are ever a part of your data flow and everything and thinking for Dr. Baker, given that they are

kind of a point of convergence of information for public and private payers, you made a comment regarding, you know, potentially skewed, you know, patient populations and everything, and I'm wondering if there's any experience or interest ever in mining state HIE data at an aggregated level to see what something closer to what might be called a truth or something, you know, that they're in that might be worthwhile. Just curious if there's any comments.

LYNNE PENBERTHY: So, I can be quick and short and then I'll turn it over. So, we've had a lot of conversations with a number of HIEs and from our perspective, it's been a little bit challenging. We have kind of dropped it as -- to some extent. However, we are having a lot of conversations with Care Quality, which does data sharing between, you know, a whole bunch of different organizations. And so, one of the things that we're talking to them about is potentially having them work with us to get some of these organizations to share, you know, common

data elements in a structured format for messages that they're already exchanging. So, I think there could be some real value there. But to try to go to individual HIEs, I think it's going to be really touch, but working through something like Care Quality might be very useful.

JUDITH BAKER: Thank you for that good question. I'm glad you brought it up. So, the answer is yes. On the sickle cell side with part of our Networking California for Sickle Cell Care, we created a data think tank. We're meeting every two months, and it's explicit purpose is to learn what each other is doing to try to synergize opportunities for our various data work on the clinical side, surveillance side, and HIE. And we do, indeed, have an HIE consultant on our team who is providing really extensive information because on the sickle cell side, and I'm sure this would be valid for hemophilia as well, but because sickle cell is so much more prevalent, we started there. What we want to frankly in the future get away with -- get away from, excuse me, is the need

for this extensive reliance on data managers and clinical research associates in the way that we currently have. We're looking for interoperability and the immediate transfer of, you know, of information across the different medical records and the different systems like Cerner, et cetera, because we have a statewide network now of sickle cell centers who are networked and we're looking for HIE solutions.

So, yes, we are engaged with HIE and we're also engaged through our HIE connection with CIEs, which is the Community Information Exchanges, and that's an infrastructure to, you know, collaborate on all of the social determinants of health. How do we increase wellness by having a more robust and interoperative organization and network of community-based organizations that provide referrals and services for housing, for homelessness, SUD?

So, we've been able to, through our HIE link, connect up to the CIE to really address

some of the social determinants of health that are particularly problematic for our sickle cell population. So, the short answer is yes, and we're really excited to see where that might take us.

JED MILLER: Thank you.

CYNTHIA POWELL: I'd like to once again thank our three speakers from this morning for their excellent presentations. Speaking for myself but also other members of our Committee, I think we feel this is really a priority or needs to be a priority for us if we're going to be able to move forward with looking at conditions that are already on the RUSP and considering new conditions for the RUSP. Tracking this information is -- is extremely important.

And anyway, with that, we're going to break. We'll break until 12:05 and reconvene then. That's 12:05 Eastern time. Thank you, everybody.

BREAK

CYNTHIA POWELL: Welcome back. For

our last panel of this meeting, we will continue the examination of key issues facing the Newborn Screening Workforce.

In May, our workforce panel speakers discussed challenges and solutions faced by the geneticists, genetic counselors, neurologists, and sickle cell provider workforces.

Today, I've invited panelists to discuss perspectives from Laboratory and Follow-up, Audiology, Pediatric Endocrinology, and Genetic Metabolic Dieticians.

Our first speaker will be Committee Member, Scott Shone, discussing the Newborn Screening Laboratory and Follow-up Workforce. Dr. Shone is the Director of the North Carolina State Laboratory of Public Health. He is board certified in high complexity clinic -- he is a board-certified High Complexity Clinical Laboratory Director trained in molecular microbiology and immunology. He spent 9 years as the Director of the Newborn Screening Laboratory for the state of New Jersey. During his tenure,

the program expanded screening from twenty to fifty-five disorders, upgraded the laboratory's Information Management System, installed and validated multiple pieces of new equipment, expanded molecular testing, increased efficiency, and reduced costs through implementation of lean processes.

I'll now turn it over to Dr. Shone.

NEWBORN SCREENING WORKFORCE: LABORATORY AND FOLLOW UP, AUDIOLOGY, PEDIATRIC ENDOCRINOLOGY, AND GENETIC METABOLIC DIETICIANS

SCOTT SHONE: Thank you, Dr. Powell. I appreciate the opportunity to speak today to the Committee, my colleagues. I actually come at this presentation today wearing three different hats. The first, is obviously as a Committee Member and sharing my thoughts and suggesting some opportunities at the end for the Committee to consider as we tackle workforce issues, specifically in the newborn screening programs, lab and follow-up. As a state public health laboratory director, from the perspective of not

only the challenges of our newborn screening program, but how they fit into the overall workforce challenges in the laboratory and broader, particularly highlighted during the pandemic response. And finally, as an elected member of the APHL Board of Directors, and how APHL's role is shaping some of the responses to this and where there are opportunities that remain. And I think it's important to say I don't speak officially on behalf of any of those three hats, but I am wearing them all simultaneously today. So, next slide, please. Next one.

So, I wanted to talk a little bit about APHL's role in developing the public health workforce and APHL recently completed a review and revisions to the strategic map, thinking about coming out, as we all plan, to come out of this pandemic in the post-pandemic area, what does the workforce look like for public health and specifically, obviously, with APHL's perspective, public health laboratories and a key component of the strategic map are to build and support a

resilient and emerging public health laboratory workforce. More than ever, the public health laboratory workforce has been completely stretched and stressed just like many sectors of this pandemic response, and we're looking at ways to broaden and support the growth of the public health laboratory including newborn screening, and ultimately, that will shape the public health lab system's role in advancing diversity, equity, and inclusion, which is an underlying goal and part of our mission as we look at expanding workforce. And that includes not only supporting lab members and recruitment and retention, but exploring new partnerships across academia and other organizations is where can we be really proactive and adaptive to the scenario we're in. So, next slide, please.

I think APHL has a history of some great programs and initiatives to approach these challenges and we'll build a foundation of where we go in the future. For example, APHL has the Emerging Leader Program, which is the 12-month

leadership development program intended to develop further leaders for public health laboratories across the country. They host a wide array of training and continuing education opportunities, have performed national assessments to look at trends that really widely affect the public health lab workforce and the most recent was published in May of 2018. So, it's published three years ago, but really the data is from the middle of the last decade, so from about 2011 to 2016. So, again, we need to refresh and look at some of that, and I'll talk more about opportunities there at the end of my presentation.

They do a lot of outreach and marketing around the promotion of public health lab science careers and provide fellowships, and I will acknowledge that I was a recipient of an emerging infectious disease fellowship from APHL coming out of graduate school, and I think -- I think serve as a model of what's the ideal outcomes for those types of fellowships as having now a leadership role for a public health lab in

this country. Next slide, please.

Specifically, about newborn screening workforce development, however, APHL has sponsored and has operated fellowship programs since 2011, so ten years now the establishment of the Ronald H. Laessig Memorial Newborn Screening Fellowship and more recently in the last couple years, the Newborn Screening Bioinformatics and Data Analytic Scholarship. The latter, obviously, is a testament to the need to assess the -- the large and diverse array of data that comes out of a newborn screening program and the ever-expanding complexity of that data. Bioinformatics needs to span the entire public health workforce, but clearly newborn screening with the types of data and volume of data we deal with, it's critical. So, that was a great addition.

And the Laessig Fellow I want to draw specific attention to for a couple of different reasons. One, two of the fellows now serve in leadership roles in newborn screening programs and newborn screening labs in this country. Dr.

Patrice Held is the co-director of Wisconsin with Committee Member Dr. Mei Baker and I had the pleasure of working with Dr. Mirian Schacter in New Jersey, who succeeded me as program manager there after I left four years ago. And so, those are great testaments to the success of these fellowships and why they are so important at bringing about -- bringing about future workforce for newborn screening. Next slide, please.

In addition, APHL, through funding from HHS, CDC, and HRSA, holds training workshops. These training workshops include molecular training for laboratorians as well as different training opportunities for laboratory and follow-up staff around tandem mass spectrometry and obviously, this was a substantial need several years ago as this technology emerged, and we need to begin thinking about new technologies that are coming out to look at new disorders and genetics and think about ways to expand opportunities for laboratorians and follow-up staff to continue to learn that, and I'll talk about why these types of

training workshops are so critical coming up.

And then with respect to mentorship on the follow-up side, the follow-up group -- well, not even a group really -- but the follow-up community associated with APHL has developed what they call FLEX, the Follow-up Learning Exchange Program, which encourages peer-to-peer connection for follow-up staff to help address opportunities and challenges that we see. And so, it's kind of a formal informal program that's had some great opportunities and met some great success at learning across the country, and there's always the informal program-to-program. I put lab-to-lab, but it's really program-to-program collaboration. Whenever anyone in the -- in the newborn screening community has a question, there are a plethora of people you can reach out to and it is by far one of the most communal and collaborative groups I have ever been -- ever worked with, and it's great to have that informal collaboration that continues, not necessarily formal, but just for the benefit of the babies.

Next slide, please.

And then identifying and seeing that there were growing and significantly emerging challenges with respect to the newborn screening workforce, APHL established a specific taskforce two years ago pre-pandemic to focus on the challenges, but obviously now having to respond to the impacts of COVID on newborn screening laboratory and follow-up staff and focusing substantially on recruitment and retention as well as to succession planning. Just like the public health workforce beyond newborn screening, the workforce is aging, and we need to make sure that we're bringing in new talent, as I mentioned earlier, with some of the fellows that have come out of the programs thus far at APHL. Next slide, please.

And so, here are some of the challenges that have been identified, not only by this taskforce, but in general, I think, by anyone who has been looking at the public health workforce and newborn screening. So, this is

where I'll put on my lab director hat for a moment and say that most of the bullets here affect my team across the board, not just my newborn screening laboratory. But there are some specific things that are crucial and hit on newborn screening specifically.

But in general across public health workforce, there are recruitment issues with respect to non-competitive salaries, particularly here in North Carolina in the research triangle, we have an overwhelming number of academic institutions as well as private laboratories, which have only grown exponentially in this pandemic that have provided substantial competitive salaries and competitive incentives as well that really is making us think about where can we -- where can we draw and where can we be more competitive on that recruitment strategy. In general, government is subject to substantial hiring freezes, particularly around budget time. When a state budget is held up, you can't move anything. And so, hiring freezes can be prolific.

There are also complex scientific and technical competencies. This affects newborn screening as well as the public health workforce, but particularly newborn screening as the need for genetic testing, whether it's first tier or second tier tests emerge beyond just kits, right? So, a lot of the disorders we're talking about right now, even drawing back on our discussions about MPS II and GAMT, these tests are not already built into FDA-cleared tests. And so, if you're going to add them as laboratory developed tests, it does require somewhat of a higher level skillset to make sure that they maintain compliance with all regulatory needs.

And I think it's important to stress, there is no specific training to come into a newborn screening laboratory or follow-up program. As Dr. Powell mentioned, my Ph.D. is in molecular microbiology and immunology and other than the molecular piece, I don't use a lot of the microbiology and immunology in my day-to-day. And so, it's really important if we don't have

pathways or identified pathways through academia that we provide that training either coming out -- coming out of school or as part of on-the-job training. And so, it's really, really important not only for laboratory but for follow-up as well. We talk a lot about genetic counselors, but not every program is ripe with genetic counselors. And so, we really need to be able to train our follow-up staff.

Dr. Freedenberg mentioned this yesterday as we had talked about adding disorders. We really need to be cognizant of the whole program and the follow-up for these disorders isn't getting any easier. And so, making sure our staff are aware of that is critical.

On the retention side, we've identified three major topics. I'm going to start at the bottom. In general, in government, there tends to be an insufficient number of job classifications or pay grade levels, which then lead to challenges in promotional opportunities or career path advancement. And then most people --

and then you have some who stay in position for a while, which is counter to my retention argument, but then that's a challenge for upward momentum. Or your upward momentum is into another laboratory whereas we might lose some newborn screening molecular scientists to our infectious disease sequencing lab because they have the skillset that can go run in there. And so, you have those issues.

And then we've had some more concerns voiced recently about personal liability. There's been some lawsuits in the newborn screening community in the recent past and beyond just lawsuits against the state and the government themselves, individual members of newborn screening programs have been the subject of lawsuits, which create substantial stress and really, you know, lead to personnel questioning, you know, I'm here to do my best job every day and this isn't a negligence-based lawsuit; it's just an argument against the way things are done and people make personal choices based on facing that

liability. So, that's something I think we need to be cognizant of as we talk about keeping up morale in our workforce. Next slide, please.

And obviously, what of the risks of having workforce challenges in newborn screening? They almost go without saying, but obviously I have a slide on them, so I'll articulate them. Low morale just exacerbates challenges. You know, it is a noble calling to work, I think, in these programs and know that you're saving babies every day. But then when you see the challenges around you, it really -- it really can tax on people and then the low morale just snowballs. And as you see colleagues leave or see the issues I've already highlighted, it really can exacerbate the problems. And that increasing need for the complex skillset we talked about has to be matched by training, fellowships, and onboarding. It ultimately comes down to competing priorities and we've talked about this a lot over the last few years as things just get piled on the newborn screening programs. I've often showed that

picture of the truck that is just piled on with people on top and top and top, and I always talk about how programs tend to be freight trains and just need help getting a car on the end of the track. But ultimately, there's a breaking point and our primary priority is always to maintain routine screening. Every day, samples come in and they have to be tested for the panel that we're currently testing and those disorders -- those results need to be reported in a timely fashion, as we've all learned. That's priority. So, what does that then jeopardize? Our disorder expansion as new disorders get added to the RUSP and added to the panels across the state and the country, those need to be validated and onboarded for the lab and follow-up teams, and then the myriad of continuous quality improvement projects that we've talked about, whether it's timeliness, cutoff assessments and evaluation. You know, dashboard development, all sorts of the different CQI activities that are necessary are often jeopardized by reductions in staff and workforce

because ultimately, testing those babies and those samples that come in every day has to happen.

So, we're often left in leadership roles having to determine where are the breaking points and what do we have to do. Next slide, please.

Okay. So, that's been a bit dire. Let me talk about some opportunities because I didn't come here just to talk about bad things but what can we do. Next slide.

So, APHL properly announced that they received a supplemental award from CDC for $27 million towards public health laboratory workforce development. I want to be clear, this is across the entire public health laboratory workforce. This money is not dedicated to newborn screening, nor is it dedicated to the whole program. This is about public health laboratory workforce development. So, APHL is going to be announcing some more initiatives as they move along and expand fellowships, which is great, but it is not the answer to what I've been talking about for the

last thirteen minutes. Next slide, please.

Really, what we need to be thinking about across the public health lab and newborn screening in particular is incentives for hiring and retention, as I mentioned earlier. I know that's sort of out of the purview of this Committee, but I do think that the more that we say it and the more venues we say it, the more likely we are to make some headway on it.

I think it's important to message the importance of newborn screening workforce. That might seem to be silly to say, but I think that during the pandemic, the focus on our virology teams and our infectious disease response and our molecular teams that are sequencing these viruses has really outpaced the focus on any part else of the public health workforce and our public health laboratories. So, really understanding that those of us in leadership need to be cognizant of the impact in newborn screening and be aware and take opportunities that are coming as a result of this pandemic to improve our entire public health labs

and our entire public health workforce including newborn screening.

I would personally love to see a coordinated approach across HHS with our partners at CDC and HRSA and NIH looking at training opportunities and coordinated training across the system and the gamut there. There is shared money, there's distinct money, and what can we do to make those resources go even further as a coordinated response, everybody in those groups sits at this table, and I think that it would be wonderful to begin to talk about how can we have a shared workforce approach to all this. And that would be to expand the training opportunities, particularly on our follow-up side and informatics but also on a high-complexity testing side and as these -- as these methods develop, really making sure that our public health labs -- not public health labs -- that our newborn screening programs and our state public health communities are the strongest and do their best job for the babies.

And ultimately, as we're thinking

about future growth, I've identified this here in North Carolina and I used to see this when I was in Jersey is you almost need a dedicated staff just thinking about what's next. I would joke that if we could just go a week without having any babies born, I just could imagine how much more we could do. I think it's up to a month now with the amount of things that are on everybody's plate. But obviously, that's never going to happen.

So, really thinking about our workforce, and not only do we need the people to do the routine testing and reporting out results and follow-up, and the CQI, but just thinking about the future and planning for that. What are the resources that we need? We spend a lot of time talking about that -- I mentioned that yesterday -- but having dedicated staff thinking about what's next would be critical to making what's next actually come to fruition even faster. Next slide, please.

So, some considerations for the future. I do think a comprehensive survey of

current staffing in newborn screening programs is critical, really taking a snapshot of what's going on. But beyond that, because we can all suffer from death by survey, that must lead to our Committee looking at those results and potentially suggesting guidelines for minimum staffing for lab and follow-up in these newborn screening programs.

The RUSP has become almost more than a recommendation in many states. We heard yesterday from the EveryLife Foundation that there are a growing number of legislatures consider auto-inclusion, whether it's two or three years. I think -- I think that we need to realize that the recommendations of this Committee sometimes take the spirit of mandate. And if we can recommend certain staffing levels, that could potentially help the programs significantly. And I would say to our advocacy partners to consider that when you're talking -- when you're using your wonderfully loud voice to advocate for the babies to also advocate for the programs to make sure that we have the resources we need. I think we

can all articulate them and having formal guidance from this Committee could go a long way.

And finally, I think those assessments should be routine. A routine check-in on how the programs are doing and a report out. We talk about status of screening. We talk about status of timeliness. We talk about status of cutoffs. But I think we need to talk about the status of the systems that support all of that and if there are challenges there, some formal recommendations coming out of this group I think would go a very long way and bear the weight of, like I said, a virtual mandate and really help us at the state level get some more work done. I believe that's my last slide.

I want to thank obviously the public health -- the Newborn Screening Workforce Taskforce with co-chair Susan Tanksley, who is our representative from APHL on this Committee. Thank you so much and everybody else pictured here. Carol Johnson -- I can't forget Carol, who has taught me so much about follow-up over the last

fifteen years and Funke Akinsola, Jelili Ojodu and Sikha Singh for helping me out thinking through these slides constructively. Thank you, Dr. Powell.

CYNTHIA POWELL: Thank you, Dr. Shone and we'll hold questions and comments until after all of the speakers have gone.

I'd like to introduce next Dr. Marcia Fort and just so nobody thinks that I'm stacking the deck with North Carolina people, we had asked the national organizations to give us names of some, you know, people who could present, and it just reflects, I think what outstanding people we have in North Carolina.

So, Dr. Fort is the Genetics and Newborn Screening Unit Manager in the Children and Youth Branch of the North Carolina Division of Public Health and serves as the North Carolina Early Hearing Detection and Intervention or EHDI Program Coordinator. She has a Master's degree in Audiology from Vanderbilt University and a Doctor of Audiology degree from Central Michigan

University.

Dr. Fort currently serves as co-President of the Directors of Speech and Hearing Programs in State Health and Welfare Agencies, DSHPSHWA. She has spent thirty-two years of experience as an audiologist with twenty-seven years of experience directly with newborn hearing screening and follow-up in public and private settings. Dr. Fort has worked with the North Carolina EHDI program since 2002 serving as Regional Audiology Consultant, Data Manager, EHDI Coordinator, and then moving into her current role as Unit Manager five years ago.

Dr. Fort.

MARCIA FORT: Thank you, Dr. Powell and thank you to the Committee for inviting us to provide some information about workforce issues, successes, and challenges to your Committee.

So, I am, as Dr. Shone said, I do wear multiple hats. I am the EHDI Coordinator for the state of North Carolina. I am also co-President of DSHPSHWA, which may be the strangest

acronym you've ever heard, and also serve as EHDI Coordinator for the state of North Carolina.

So, today I am presenting information on behalf of EHDI programs across the country representing the DSHPSHWA organization. Next slide, please.

So, this is a quick slide. I'm going to go into a little more detail about each of these workforce challenges. Just the scope of the EHDI program, funding and sustainability, incongruent policies and/or regulations, diversity of skills and stakeholders, shortage of qualified professionals, insufficient enforcement ability, benchmarks that are dependent upon others, turnover and institutional knowledge, and mentoring. Next slide, please.

Uh-oh, that shouldn't be my next slide. Okay. I had -- just go back -- if you could go back, please. Go back a slide. We'll stay on that slide. I have some additional information and have some updated slides that I will provide to you.

So, talking about the scope of the EHDI program, it's just a reminder for everybody on this Committee that EHDI has only been a national screening and follow-up program for twenty years. So, we're -- we are still very young as a national program and have come a very long way in a very short period of time.

So, the original scope of early hearing detection and intervention twenty years ago was newborn hearing screening, diagnostic audiology evaluation, referral into early intervention, and aggregate data reporting once annually that included fifteen data items. Next slide, please.

So, twenty years later, this is the scope of the EHDI program, what we are required to do. It's newborn hearing screening, and we are required to provide follow-up not only for abnormal screenings, but also for all infants who did not have a screening at their birth facility and any infants for which there is no documented hearing screening reported to the EHDI program.

So, our follow-up requirements do expand greatly beyond just abnormal screening.

Diagnostic audiology evaluation, enrollment into early intervention, family engagement, deaf mentoring -- we've got deaf and hard-of-hearing adult involvement with families of newly identified children with hearing impairment -- health information technology, electronic data system development and integration with other programs such as state labs, vital records, and early intervention, late onset hearing loss -- so, now they've moved us beyond newborn hearing screening -- early childhood hearing screening up to the age of 3 years, cytomegalovirus education and outreach, and our data reporting has now moved to de-identified individualized data reporting for each and every birth in our jurisdiction with over 170 data items required on each birth and we report that at least twice annually.

So, in the course of twenty years, the scope of the program has grown dramatically and, as we'll see going forward, funding did not

grow proportionately with that. I will say that most -- a lot of EHDI programs do not have more than maybe two or three FTEs that are expected to carry out all of this work in their state. North Carolina is fortunate in that we -- we do have access to a larger staff. But many states only have one or two. Next slide, please.

The funding for EHDI programs is extremely limited and there are concerns about sustainability. So, fifty-nine states and territories receive $235,000 a year from the Health Resources Administration. That is $235,000 per year, per state, regardless of the size of the birth cohort. Every state and territory receives the same amount of funding. Thirty-nine states and territories currently receive funding from CDC. The cap on that funding is $160,000 per year. Next slide, please.

So, DSHPSHWA conducted a sustainability survey among our membership in 2019 and '20. And so, this data comes from that survey, and we had forty-eight out of fifty states

who participate and responded in this survey. 75 percent of our respondents do have some legislation governing EHDI, but only 14 percent of these included any funding or budget note. So, 76 percent of our responding states were unfunded mandates. 30 percent of the responding states have some contribution from the state general funds. Again, 70 percent have no state general funds or are not mentioned in the state budget. 27 percent of our responding states receive a portion of the newborn screening fee. Again, I flip that around to say that 63 percent do not receive any funding from the newborn screening fee. 52 percent of our responding states did say they have access to Title V funds but only 21 percent of those stated that they feel like that funding is reliable and in many cases, it's only available for things like travel to conference but not for staffing. Next slide, please.

Other workforce challenges; incongruent policies or regulation. So, we are dealing within EHDI because we are required to do

the screening and the follow-up and the diagnostic, that is governed by health care, but we are also required to ensure enrollment into early intervention, which is governed by educational policy. So, we have constant concerns with HIPAA and FERPA and privacy regulations that don't necessarily support one another. Federal program authorizations don't always support one another and the example I will give of that is the EHDI Reauthorization through Congress mandates EHDI programs report out early intervention, enrollment, and outcome data, but the IDEA Federal Authorization that OSEP comes down through OSEP and Part C does not have a similar requirement for cooperation and without those policies and agencies working together, it makes it very difficult when one party's funding is dependent upon successful execution and the other party's participation is not supported through their authorization or regulations.

We also have, as Scott mentioned, sometimes policy statements or position statements

are viewed as the gold standard and requirable and we have that with the Joint Committee on Infant Hearing Regulations. But we have conflicting information with that in that JCIH recommends that EHDI programs are led by audiologists and then deaf consumer organizations, deaf and hard-of-hearing organizations insist that EHDI programs must be led by deaf and hard-of-hearing individuals, and then we have family-based organizations -- if you remember back to my statements on the scope of work -- we're required to have family engagement throughout all of our work and our family-based organizations and parent support groups want -- they feel like EHDI programs should be led by families of children who are deaf and hard-of-hearing. So, we have some incongruencies that we are trying to navigate on a day-to-day basis.

The diversity of skills, again I go back to many of our EHDI programs only have one or two or three individuals who are trying to accomplish all of the required tasks. Though we

need our workforce to have so much diversity in their skillset, program management, program evaluation, quality improvement, IT programming, health information technology, outreach, hospital regulations, audiology regulations, speech and language and hearing normal development, educational policy, communication options and access, hearing technology, hearing aids, cochlear implants, grant writing, contract development and monitoring, and all with a very, very small workforce.

And the stakeholders that we are required to have on our advisory committees and work with on a regular basis include audiology, newborn screening, laboratory, Medicaid and other pay sources, early intervention, deaf and hard-of-hearing adults, lend programs, parent support organizations, hospitals, genetic counseling, home visiting, WIC, vital records, physicians from ENT, pediatrics, family medicine, and genetics, graduate training programs, schools for the deaf and deaf/blind among others.

So, another challenge that we have with our workforce is a shortage of qualified professionals. Again, as Dr. Shone mentioned, there's no -- I believe he mentioned no specific training or designation for newborn screening staff. The same thing exists for audiology. There are -- there is no specialty certification or differentiation for pediatric audiology and there -- it requires a different skillset for pediatrics, which encompasses a wide age range and what is needed to diagnose and treat infants for hearing loss. So, there's a shortage of professionals that really have the skills and knowledge and equipment and needs to work with the infant population. There's also a shortage of qualified professionals as teachers of the deaf, interventionists for the deaf, ASL interpreters, speech translators and others. So, we're dealing with a shortage of professionals in a variety of realm. Next slide, please.

Another challenge that we have is insufficient enforcement ability. Many of our

states, we do have legislation that mandates that there be a program, but we have -- do not have any teeth, so to speak, to enforce when things don't go as planned. So, we have a hard time actually enforcing that procedures are followed as we would like at all of the birthing facilities and midwives and outside agencies that we're so dependent on.

Our benchmarks -- so, we want everybody -- every baby screened for hearing loss by 1 month of age. If they don't pass that screening, we want them diagnosed with their hearing loss by 3 months of age and we want them enrolled in early intervention by 6 months of age. All of those benchmarks on which our limited federal funding are dependent on really rely on other professionals to do what we need them to do. We have to be able to get appointments. We have to be able to find transportation. We have to have referring physicians that recognize the importance and the urgency of following up on a failed hearing screening in the way that they

recognize the urgency of following up on a failed newborn metabolic screen, and there is not the same sense of urgency with those professionals.

So, being so dependent for program success on other people is difficult. So, all of the challenges that I have mentioned so far lead to a very high stress level among EHDI program staff and significant amount of turnover just within -- since February of 2019, twenty-one states and territories have had turnover in their EHDI coordinator just since February of 2019. Two of those states have turned over the EHDI coordinator three times in that period of time and two of those states -- three of those states have turned over their EHDI coordinator two times in that period, and this is -- has been fairly typical of our workforce for the past several years.

We also have to deal with the turnover in hospital screening staff -- the people that are actually completing those hearing screenings in the hospitals and there's frequently

turnover there as well as other staff besides the coordinator for state EHDI programs.

Then, I want to talk a little about mentoring. We do have a very strong EHDI community nationally. We support each other very well on a national level. We do have a mentoring program that DSHPSHWA is running for new EHDI coordinators. The challenges come because we can -- we can guide and mentor pretty easily on the national requirements and grant requirements and help there, but once you get into the state level, every state's bureaucracy or methods of doing things like approvals for grants and purchasing and contracting are very, very different, and because of the small size of our programs, there just aren't people who can provide that mentoring that is direct to EHDI. So, it becomes very difficult there. And then also mentoring at the hospitals and audiology practices when that turns over. Next slide, please.

So, again, like Dr. Shone before me, we don't want to talk just about our challenges

but also some potential solutions.

So, we would like to see some improvements in sustainable funding. Again, I think you saw how limited our funding is compared to the scope of what we do, and I'm going to move this into the next bullet which is continued/increased collaborations. I think support from Committees like yours could potentially assist us in getting some funding through newborn screening fees, which are much more stable than grant funding or Title V funding or insurance-type funding. The continued and increased collaborations, because of our very limited funding and because of the scope of our work, we have, as national unit of EDHI, we have been very creative and very collaborative with many partners and have some really strong collaborations with HRSA and CDC and APHL and the Joint Committee on Infant Hearing and Lend programs and audiology training programs and the AAP. So, we have some really strong collaborations. We need to definitely continue

those and then increase them in the areas where we've not been as strong and can work together to really strengthen all of the needed workforce issues with our groups that work with these infants and young children.

And then an improved sense of urgency. It really is a challenge for us that physicians do not see the urgency of following up on a failed newborn hearing screening in the same way that they do follow-up for abnormal metabolic screening, and we frequently deal with families who are told by their physician, oh, it won't matter, just wait. They're not old enough to test. It will be fine, we'll check back when they're 2. So, if we could collectively work on improving that sense of urgency, I think that would -- that would be helpful.

And I believe that was my last slide. So, I want to thank you again for giving us the opportunity to present to you today and look forward to working with you in the future.

CYNTHIA POWELL: Thank you, Dr. Fort,

for your very informative presentation.

Our next presenter is Dr. David Allen. Dr. Allen is Professor of Pediatrics at the University of Wisconsin, School of Medicine and Public Health and Head of Endocrinology and Diabetes and Director of the Endocrinology and Diabetes Fellowship Program at the University of Wisconsin, American Family Children's Hospital in Madison.

On the national level, Dr. Allen has formerly served as Director and then President of the Pediatric Endocrine Society. He served as chair of the Wisconsin Endocrine Newborn Screening Committee from 1991 to 2015 and as member of the American Board of Pediatric Endocrinology from 2010 to 2015.

I'll now turn it over to Dr. Allen.

DAVID ALLEN: I'm not unmuted. Yeah, thank you. I want to thank the Committee for this opportunity to present and be a part of this very interesting meeting as a guest. You know, some of my credentials were mentioned there and most

recently, I also served as the chair of the Pediatric Endocrine Society Taskforce looking at workforce issues and I'll be presenting some of that data today. I want to make a special shoutout to Shawn McCandless, who I saw on the member list today. Shawn and I shared some residency time together in Madison. So, it was great to see him again. Next slide, please.

So, it's been my privilege over the course of my career to really work with the Wisconsin Newborn Screening Program, really one of the innovative and forward-thinking programs that I think has kept us on the cutting edge. You can see I've highlighted the endocrine disorders there that are screened for -- congenital adrenal hyperplasia and congenital hypothyroidism -- and I guess one of my underlying messages today is, you know, this has been fairly stable. We've been screening for the same disorders for the last thirty years, and it's really what else is going on in our subspecialty that is really influencing our ability and threatening our ability to care

for these children rather than an expansion in the screening programs themselves. Next slide, please.

So, just by way of review, this is our algorithm for screening for congenital hypothyroidism. You can see this frequency is about 1 out of 2,000. It's interesting over the course of my career this frequency of diagnosis has just about doubled for a variety of reasons. It's obviously a very important disorder to screen for, and our state like most, but not all states, uses a primary TSH approach to detect severe primary hypothyroidism primarily. The TSH is dramatically influenced by the timing of collection, and I'm proud to say that, you know, the Wisconsin program really pioneered the development of eight specific cutoffs for TSH criteria, which made a substantial improvement in the false positive rate.

These children come in all varieties of severity. We are very cautious about implementing treatment whenever there's a question

about possible congenital hypothyroidism, and we usually are able to determine the permanence of the need for thyroxine supplementation by 3 years of age. I put the therapeutic objectives there for you.

The pediatric endocrinologists are, of course, critical for follow-up, particularly in the first five years where, you know, the central nervous system development is so dependent on adequate thyroxine. So, these children are getting laboratory studies every three months or so in the first two years of life with very frequent dosage adjustments to keep free T4 and TSH levels in the target range. A couple of visits with the pediatric endocrinologist, but really the laboratory studies are paramount. I'll come back to this message later, but this is one disorder where the advent of telemedicine and its expansion has been very helpful and it's quite adaptable for the follow-up of this disorder. Next slide, please.

Now, when it comes to congenital

adrenal hyperplasia, this is a bit more complicated. Again, this is an autosomal recessive disorder with that frequency that I mentioned or show there. You know, these screening programs are primarily designed to detect 21 hydroxylase deficiency, but because of the pathways that are involved with other forms, it does detect some of the -- most of the less common forms of congenital adrenal hyperplasia as well. As this Committee probably well knows, this is a defect in cortisol synthesis that ties these disorders today. So, the risk of undiagnosis is cortisol deficiencies plus/minus some degree of aldosterone deficiency and, of course, the side effects of the adrenal androgen production virilizing female genitalia and the central nervous system.

You know, before newborn screening, this was -- could be a fatal disease, especially in the young recognized males and some of the females also experienced sex-misassignment. The screening is currently and still relied on the 17

hydroxyprogesterone, which is the metabolite that accumulates before the 21 hydroxylase block. This is very influenced by gestational age and birth weight. Again, one of the pioneering studies in Wisconsin was to determine cutoffs that were based on birth weight, and this has really been adopted across the country and also throughout the world to limit the false positive testing. But there still is a significant problem with false positives, and that's where now the mass spec is providing some valuable initiatives. And I heard Patrice Held's name was mentioned earlier. She and I are collaborating on looking at some of the other metabolites that can improve newborn screening using that second tier testing.

The treatment, of course, is lifesaving for these children with cortisol and mineralocorticoid replacement and this is a very delicate disorder to manage because the growth -- the normal growth of the children is really dependent on the appropriate balance between control of the disease without growth suppression

by the glucocorticoids. In addition, especially early in life, there are many illnesses that require stress dosing, lots of calls to the nursing team and so on and so forth, and then in contrast to congenital hypothyroidism where the management really simplifies as the children get older, in many ways, the congenital adrenal hyperplasia management really becomes more complex as the children go through puberty and then there's a lot of issues that overlap with the psychosexual development as well as their medical management. Next slide, please.

So, how are we doing with regard to the ability of pediatric endocrinologists to care for these children? Well, there is a shortage of pediatric endocrinologists, and here's some data from Wisconsin just showing how far the average child has to travel to get to the endocrinology specialist, you know, thirty miles on average, but many kids are traveling two to even three hours to get to a pediatric endocrinologist and you can see that it's harder to get to the pediatric

endocrinologist than it is to many other specialties, for whom the patient population is quite a bit less. And in the country, there are ten states that have fewer than one pediatric endocrinologist for every 100,000 children. Next slide.

And, you know, unfortunately, the trajectory of the pediatric endocrinology workforce is going in the wrong direction and that is really what prompted our society to look into this in depth and try to make a diagnosis of the problem and make some recommendations.

The recruitment has been very problematic over the last ten years where the number of endocrine fellows is actually declining substantially from 2012 to 2018, and we're currently experiencing an applicant to position ratio of 0.7 so that in our last match for fellowships, you know, not quite half but, you know, 40 percent of the positions went unfilled. Next slide, please.

And I show this graph, and I just

call your attention to the two graphs that are highlighted in red basically showing an increase in the number of unfilled positions after the match, and again, this is not only through the match but even after recruiting foreign medical graduates to try to fill positions. You know, we still have thirty positions that are not being filled and more dramatically, the number of unfilled programs going up from eighteen in 2014 to twenty-nine and I think it was even in the thirties here with the 220 match. So, many, many fellowship programs are not able to attract candidates. Next slide, please.

And in addition to the dwindling recruitment, you know, there are some other challenges that limit the ability of the current workforce to provide the patient care and that is that because of the diminished recruitment, we are an aging workforce. We have a fifth of our group that is over 60 years of age, many of whom are working full-time are not seeing a full clinical panel anymore, and interestingly, you know, we're

now attracting at least 80 percent women into our ranks in their early career, and many of these women, for obvious reasons, are working part-time so that the numbers of pediatric endocrinologists do not really represent the FTE that's available to provide clinical care. And we continue to struggle with underrepresentation of minorities. We do have 23 percent of the trainees that comprise this group but only 5 percent of the current workforce is Black. Next slide, please.

And on top of this, as I mentioned at the beginning, you know, what's really pressuring the pediatric endocrinology workforce is the growing patient population that we're experiencing in other things that we do and this is most dramatic with the diabetes where not only the Type 1 numbers have grown tremendously, but especially the Type 2 diabetes, which has just exploded over the last fifteen years and again, all the other obesity morbidities that accompany it. And in addition, there's been a number of other disorders that have become much more a part of our practice

-- transgender medicine, cancer survivors, and also retaining these complex patients well into their 20s before they get transitioned to adult. So, we have these synchronous trends where there is declining recruitment and increasing patient numbers, which is putting a tremendous strain on the number of endocrinologists available to follow those kids diagnosed by newborn screening. Next slide, please.

So, why is this happening, you know, why are we having such a difficult time maintaining a workforce pipeline? Well, there's a number of factors that we identified. One is that, you know, the critical time for medical trainees to make their career decisions is usually in the last year of medical school or maybe the very early part of residency. By the time they're in their second year of residency, people have differentiated. And the problem with a number of specialties is that they don't -- just don't exposure during medical school rotations and also during residency. It's much more common to have

experience with these fellowships during the third year, long after somebody has made their decision about where they're going to go with their career.

Financial concerns are a major issue. I point out there the average medical student is almost a quarter of a million dollars in debt at the end of their education. So, the idea of deferring salary increases for an additional three years of training is a powerful disincentive and, of course, when it comes to pediatric endocrinology -- and I'll show some data later -- this is coupled with a relatively lower average salary compared to other areas of pediatrics.

And I also think that, you know, in the last several years, we are facing some headwinds with regards to people's perceptions of quality of life as it relates to pediatric endocrinology, particularly with the burden of still providing pretty much continuous availability to our diabetes patients and overnight call and weekend call and that sort of thing. So, the boundaries between personal and

professional life are really considered to be somewhat unpredictable for the pediatric endocrinologist, and we're facing a lot of competition from other specialties, which are being organized really along shift schedules, and I especially think hospital medicine is putting a dent in academic specialists, especially nonprocedural ones, because there's a lot of overlap in the candidates that we otherwise would attract. Next slide, please.

And I just wanted to show this graphic about the dramatic effect of differences in financial earnings over the lifetime, which, you know, these graduates or these medical students are keeping an eye on this. So, this is recent data that was published in Pediatrics just showing the difference in lifetime earnings and you can see general pediatrics there, fourth from the left, is kind of like the baseline and you can see most specialists make less over the course of their lifetime, but it's especially profound when it comes to what we call nonprocedural specialists

and I highlighted endocrinology there with a lifetime earning that's about $1.5 million dollars less than that of a general pediatrician. So, this is a person who does no additional training after residency and, you know, of course when residents are looking at this and weighing the risk, the costs, and the benefits of doing additional training, this does not look very attractive. Next slide, please.

And when it comes to pediatric endocrinology, unfortunately, this is a comparison between how things looked in 2010 compared to how they look now, and the deficit that we have accumulated even in addition to that over the last ten years has been the greatest of any of the specialties, and you can see on the left side of the diagram the procedure-oriented specialties have had more gains and the cognitive specialists or nonprocedural specialists have had greater losses over the last ten years. So, this is profoundly affecting recruitment to our and some of the other nonprocedural specialists. Next

slide, please.

And this, of course, has a dramatic effect on how many people are going into the specialty and how hard it is for patients to access these, and on this graph, you basically see this relationship between lifetime earnings and the specialist to child ratio with these, again, these cognitive specialties being concentrated on the left side and the point I would make for endocrinology is that the number of patients within that population that have endocrinology problems -- diabetes, obesity, and what not -- is far in excess of what you would see compared to infectious disease, nephrology, or rheumatology. So, the number of endocrinologists that are actually available for a child that would need a pediatric endocrinologist is a tremendous outlier compared to even this graph as shown. Next slide, please.

So, what's needed to change this trajectory and, you know, restore an adequate supply into our workforce? Well, we feel strongly

that, you know, we need to have more exposure to people earlier in their training so that we can show them the positive aspects of being a pediatric endocrinologist. So, we're lobbying very hard to have outpatient specialties be a part of core rotations during medical school. We really want to get these people in front of enthusiastic mentors. We're working with a variety of organizations to influence residency training and really make exposure to outpatient specialties a part of the intern year and not to defer it until later and also to get the professional societies really involved in contacting medical students and generating interest amongst medical students at that point in their career. Next slide, please.

But we have to also address some of the barriers, I think, in terms of while we also try to make the specialty look more attractive or get earlier exposure, and a major part of this is financial and this is, of course, an area where I think committees like this can add their voice to

the lobbying that's needed to make people feel like they're going to be able to pursue these interests without experiencing significant financial setbacks. And so, expanding loan forgiveness for work in underserved areas, that would be very helpful. In particular, we want to see a funding of a targeted loan repayment program. One of my concerns is that if we just fund the, you know, if the loan repayment program in general just gets funds for all specialists in pediatrics, that's really not going to help the nonprocedural specialties to the degree that we need. And obviously, when it comes to reimbursement, it's important that we continue the movement toward valuing the input the nonprocedural specialists bring to the table. There has been some movement in that direction with regard to time-based billing, which has been helpful in the last six months, and we just hope this momentum can continue. Next slide please.

We are continuing to reevaluate whether or not it would be of value to think about

generating two-year program trainings to limit some of the loss financially. This is the way adult medicine approaches most of its fellowships. Pediatrics remains an outlier. But there still are substantial barriers to making that change when it comes to adequately training people for research careers and so on and so forth. So, it's not clear at all that changing to a two-year program would really modify workforce.

And also, to pay attention to some of the perceived lifestyle detractors. We want to find ways to expand utilization of care extenders, particularly to influence the care of the diabetes population so that this isn't falling all on the physicians and also embracing technology, which can improve the work and personal life balance and I think there are some particular disorders in endocrinology, which can lend themselves to this work. Next slide, please.

And I'm happy to see that this Association of Medical School Pediatric Department Chairs has started this Pediatrics 2025 Initiative

that really is looking at all of these areas that I've discussed and really trying to change the medical education paradigm, get exposure of these specialties to medical students during their critical career decision years, and also to really address some of the economics. So, this -- this is a concerted effort, which I hope will bear us some fruit in the next four or five years. Next slide, please.

So, I just wanted to close with, you know, some of the solutions, I think, or the needs that are present to really ensure the optimal follow-up of children that are diagnosed with these endocrine disorders by newborn screening. I can't emphasize enough how much it is -- how important it is for newborn screening programs to be in close collaboration with the specialists that they have in their state, and I think the Wisconsin program has just been a paradigm of how this can be done effectively and that is the first and critical step, I think, to really ensuring that these children are followed up adequately.

As I pointed out, you know, I think to keep up with these -- this work demand, we're going to have to find ways of recruiting more pediatric endocrinologists to really keep the specialty viable.

We can use the systems from care providers in terms of physician's assistants and nurse practitioners. I think this can be effective in general hypothyroidism much more than it is for congenital adrenal hyperplasia. There's no question that the complexities of managing congenital adrenal hyperplasia really demand that depth of knowledge that only a fellowship-trained expert can provide, and I think also there's limited ability of academic institutions, which are where most of the pediatric endocrinologists are, to really offload a lot of this work to nurse practitioners, mainly because of limited funding to fund these positions.

You know, in some situations, adult medicine collaboration can work well. Again, I think this can be okay for congenital

hypothyroidism where either family practitioners or adult endocrinologists are very capable, of course, of managing thyroid replacement in adolescents and beyond. Again, it's been a very -- it's been very problematic trying to transition a congenital adrenal hyperplasia population during these years because there -- there is so much going on with them critically, psychosexually, as well as medically during their adolescent years that transition to adult medicine seems to be a significant problem and most of these kids are followed into their early 20s by pediatric endocrinologists.

And the technology, I think, can be part of the answer. It does improve patient access. I'm currently doing about 50 percent of my congenital hypothyroidism follow-ups by telemedicine. It's a wonderful thing for both the patients and for our -- the burdens on our clinical access. So, I think that's a definite step forward. But importantly, I don't think we can let the payers believe that this is going to

diminish the shortage of the providers because whether I'm seeing them by telemedicine or in person, it -- it doesn't change the number of providers that are needed to provide that service.

So, with that, I'd like to thank you again for your attention. I look forward to any questions that there might be at the end of the session. Thank you, Cynthia.

CYNTHIA POWELL: Thank you very much, Dr. Allen, for that excellent presentation and something that those of us in pediatric genetics and metabolism certainly can relate to.

Our last presenter for this panel is Dr. Rani Singh speaking on behalf of Genetic Metabolic Dieticians International. Dr. Singh's research career focuses primarily on the study of intermediary metabolism and translating this discipline into genetic nutrition for children with rare inherited diseases. In addition, she serves as the PI for the Southeast Regional Genetics Network, SRGN, funded through HRSA. Her research focuses on optimizing the nutrition

treatment of genetic disorders by investigating both clinical and biochemical health markers, evaluating the efficacy of restrictive diets and genotype/phenotype relationships in inherited metabolic disorders while developing patient education and community outreach strategies.

I'll now turn things over to Dr. Singh.

RANI SINGH: Thank you, Dr. Powell. Needless to say, we at GMDI were totally thrilled and thankful for the invitation because we do know how important the support from this Committee to the field of genetics nutrition has been. The Committees call in support of important letter of support and publications in 2020 for stable and affordable access to medical foods was the testimony for acknowledging the role of nutrition in this area. And while we continue to tackle that issue in medical foods, I think the topic of a trained workforce is so critically related in the field for optimizing the care and improving outcomes for the patients in the patient

population of inherited metabolic disorders. Next, please.

So, today I'm going to present on behalf of GMDI the important role for genetic metabolic dieticians play in newborn screening and long-term follow-up, some emerging activities in this field and to address the needs of current workforce and challenges and some future needs and plans. Next, please.

So, we -- in 2005, the expansion started happening in metabolic disorders due to the technology, we felt that there was a need to develop specialized skills for RDNs in this expanding field. We needed an infrastructure to support research and training of the RDNs so they can easily manage these disorders requiring complex nutritional management.

GMDI was founded in 2005 with a very small intervention grant from Emory University with a handful of committed founders with a very clear mission to provide standards of excellence and leadership in nutrition therapy for genetic

metabolic disorders through clinical practice, education, advocacy, and research.

We have since then successfully built a membership of close to five hundred members with great international interest. We can boost about our successful meetings, which bring members and talkers together for new knowledge and training for continuing education. We have had an average attendance of about over four hundred attendees at the recent meetings. In addition to the training, we have developed the first ECHO program for genetic nutrition training that have been developed in partnership with SERN. In addition to GMDI and SERN collaborations, I think we have very smartly collaborated to lead best practices -- to develop best practices through our Guidelines Project, which is not only made available to open access to web-based programs globally but has also been published by peer-view journals and I -- and I think that has given a lot of infrastructure to start our practice in this newly emerging field.

And we also collaborated on advisory

boards with other organization agencies like SIMD, Academy of Dietetics, parent organizations, and really bring in very specific perspectives about the care needed in this area. Next, please.

So, a very basic reminder that this paper by Dr. Brad Therrell highlighted the importance of nutritional intervention as the primary therapy in all these disorders on the RUSP panel, which are bolded indicate that in order to achieve really good outcomes, immediate nutrition intervention is necessary in these disorders. Next, please.

So, also the Secretary defined the goal of long-term follow-up as assuring the best possible outcome for individuals with disorders identified through newborn screening, and we at SERN have done a paper with Dr. Alan Hinman in Public Health Informatics Institute mapping this process after the blood spot screening and yesterday we know a lot of discussion occurred. It's a system and it doesn't just have to stop with the screening, and this was an attempt at how

the system can be integrated with technology at that time and the steps were shown sequentially.

As you can see why we move in the direction towards the diagnosis and treatment being moved from a public health-based program, which is the screening program -- screening program grounded in public health towards more insurance-based care, which brings challenges and barriers including the reimbursement issues of the nutrition services.

So, while traditionally we have thought about treatment and management after diagnosis, I kind of want to share my personal experience. I feel that the road starts right after screening in many cases in a child because we know in order to have good outcomes, because we know the child has to be fed appropriately while we are still awaiting for the confirmatory testing, which can take a little time, and -- and I think we have an opportunity to -- if we understand the new technology of tandem mass spec, metabolomics and the interpretation, many times we

can start intervening with a cautious approach while awaiting for those results and then, of course, the important role for dietician of going management and treatment with lifelong monitoring, and it's very interesting to me because up until now, I've been doing the care for the PKU patients for twenty-seven years personally, and now I -- in the last three years, I've had women in 50s coming in and wanting to talk about menopause and how does the genetic disease picked up on newborn screening affect the late stages in the elderly, which I'd never personally thought I would get to witness. We were always thinking about as newborns. So, the transitioning issues and getting interventional help with proper nutrition.

So, a dietician has to be confident, knowledgeable, and needs support of their team both from the public health end and the clinical team. This is an evolving game, you know, skills for lifelong nutrition follow-up, breadth of knowledge through life cycle is so necessary. So, we are talking with not just a pediatric dietician

or neonatology, somebody who understands genetics and nutrition through life cycles. So, we do need to build evidence for our interventions and I want to end this slide by saying that metabolic RDNs can play a very important role with long-term follow-up for disorders to inform evidence-based practice, and it's a huge opportunity for us to collect point-of-care nutrition data to inform and build evidence-based interventions and personalize it further and not just based on book knowledge and theoretical principles. So, next please.

So, the critical role for successful management, as I mentioned, starts with immediate initiation of treatment to prevent intellectual disability, crisis, and even death in some cases, and that is highlighted through our guidelines in AAP and newborn screening guidelines, NIH recommendations, health people, et cetera.

But also, we know that the Committee had defined the long-term follow-up goals around four major components that were identified and dieticians played a role at every level. The care

coordination starts immediately for these metabolic disorders right away to have access to necessary formula to feed the baby, like I said, in the right manner. The evidence-based interventions to ensure that there is access to medical food, medications, and lifelong care, and the knowledgeable dietician.

And I think it's very important that laboratorians talk about, that we continue to see what works. How can we improve? Because even like with a big practice like the academic institution where I work, the clinicians can have different ideas on how a patient could be treated. We really need to continue to have this dialogue, not only among us, but nationally. What is working, what is not working and the knowledge generation, collecting and documenting data for clinical trials and registries as we work with these patients and we talk about putting proper information in the electronic records and collecting the data at point-of-care can be such a huge contribution.

So, I do think that the trained workforce is needed and the role of the trained workforce is to offer medical nutrition therapy and create a nutrition therapy plan that serves as a guide for treatment, the care plan that offers a collaborative approach between patients and their support system. When I talk about social support system, I'm talking about the families, the schools, the foster homes, the nursing homes to maximize the wellness for mind and body. And these specialized food-based dietary recommendations and nutritional supplementations to correct the deficiencies to enhance the pathways as well as education and resources for patients, families, and caregivers are all common activities needed by a trained dietitian.

In addition, not only in the outpatient setting, we need to set up all these networks to have quick access, and I think we can create national networks and communication tools, and resources so that they can be accessed easily and in a more harmonized way. But also to help

with the inpatient admissions for the nutritional care. I cannot tell you let's take an example of MSUD. We can manage outpatient great. But when they are sick and they go to the hospital and are not managed appropriately with a good nutritionist, I can tell you, we can tell you we change the life path of the child. So, I think it's very important for us to think how this happens after screening, how the system works after screening and how to integrate that. Next, please.

So, I also feel that having worked in the field for over thirty years, we have seen the image for dieticians from the kitchen to learning and bringing the sciences. We look at the metabolites, we look at the genetics already, and this is a huge opportunity and then we take the nutrients to special medical foods, for protein modified foods, and the sources of intact protein and we modify the diets around the knowledge of metabolome and the genome and I think the important opportunity that we can lead the area in

emerging precision nutrition field by furthering the knowledge through research with probing through microbiome and exosome. We talked about social determinants and capturing all the data in this area for nutrition and taking it to the next level of precision nutrition to generate new knowledge and really inform our current practice. Next, please. Next, please. Next slide.

So, the roles and responsibilities of the dieticians can vary based on the area, and it's amazing that in this area now, we have an opportunity to work in the clinical settings and public health settings, on state newborn screening advisory boards, work as researchers in clinical trials and patient registries, independent researchers. Several of us are doing industry-sponsored investigator-initiated protocols, which support research, serving in educational -- in academia and medicine in universities and industry. We have a large workforce, which has been recruiting by industry in several different roles as researchers, medical science liaisons,

sales and marketing, and then, of course, with the government, opportunities to work in this area are emerging. Next, please.

So, based on the survey done by the GMDI, we recognize that the RDNs are specializing -- who are specializing in this area can work in a variety of public and private sectors. Most of them, 56 percent, are working in university medical centers, 20 percent in public hospitals and medical facilities, 12 percent in private facilities, and a large number, 20 percent of our list serv members are with industry. We have been told we are competing for salaries with them. They are given much better opportunities to not only make more money but also advance in the field.

So, the way the funding is going, what we understand, most of the hospitals, fee for service and they're salaried, there are some state health departments which are including some dietician support and some newborn screening contracts have, it's piecemeal, and most of them

in outpatient are through fees for multidisciplinary team visits and they are being billed usually under a bundle or a physician rather than as independently. There are centers billing for independent, but the fees come out of that funding to support the salaries. Next, please.

So, the dieticians who are specializing in working with individuals in this field, they require a unique skillset in contrast with other clinical specialty areas. The specialized training is necessary for the dieticians choosing to work with this complex population, and there's currently no requirements or recognized training for the RDN working in this field. So, we definitely need to start thinking how to standardize that in this expanding field and really address the unmet needs for all the patient education and nutrition follow-up that is needed, which we feel can prevent a lot of hospitalization for the patients.

Dieticians are overburdened with the

care coordination component because they cannot do their job if the patient doesn't even have access to the medical food or the low protein modified foods that are needed to manage them. There needs to be a clarity in roles. We have started that work with GMDI and published a paper of the core competencies that are required and unique in this area. We need to figure out a way, where do they go in the clinical setting other than just calculating diets and then how do we retain and promote and give them opportunities as part of the genetics team, things to grow and bring the leadership skills, and how can we allow more opportunities for them to see the patients independently if they are licensed and registered in the state? So, general message from the surveys we have received is that they are very much underpaid and overworked. Next, please.

So, the data which we looked at reflected that on average, there's one dietician for one hundred and thirty-three patients management -- this complex management, and the

disparity between earnings and responsibilities is tremendous. This is after five years of employment, the metabolic dietician average salary in 2020 was very compatible to what's shown by academy, around $70,000, but the starting salary of your post-master scholarship in Southeast Region is around 55,000. And this is after a master's and two years of fellowship is what I'm faced with at my institution currently. The genetic counselors in 2020 were making close to the average salary, but there's no standard certification or credentialing, and that's something that we definitely need to further look.

There's uneven geographic representation. We have some institutions that have -- in big academic institutions, we see multiple dieticians, whereas in others, we see maybe one covering even three clinics. Limited diversity within the workforce. We have inadequate reimbursement for the medical nutrition therapy services and the time spent, our survey showed, that the average dietician is spending

more than five hours per week just on prior authorizations and writing letters of support to the -- to the insurance companies for advocacy for the treatments that they need. Next, please.

So, where do we go from here? I feel that we do need to figure out a way to support nutrition services and medical nutrition therapy to individuals with genetic metabolic disorders, particularly with the shortage of medical geneticists. I think giving detailed diet is what is going to improve the outcomes in these patients and we need knowledgeable people doing that. We need to enhance the diversity of the nutrition workforce. We need -- I think one of the things we learned during COVID, particularly genetic nutrition can really take advantage of telemedicine support and increase management because they don't need to wait six months to come to see a clinic and a medical geneticist. They need to be monitored monthly to adjust for their weight and all early on in the newborn periods or during pregnancy to be able to prevent

hospitalizations and really have good outcomes. And this -- we can be in their homes, in their kitchens, helping them make their food into their nutrition therapy, so it's really critical we consider some public health models to have access to a trained workforce.

Add access to medical foods. I would recommend that we should consider when we are thinking of developing newborn screening programs -- and this is a global issue -- that we do add access to genetic metabolic dietician and medical food as a quality indicator of newborn screening programs. I think it's important not to see that as an afterthought to say we are going to start screening. I get from other countries who are trying to do this in developing countries and really pushing them to have this dialogue up front in the program planning phase and to increase -- I would encourage that as we think about long-term follow-up patient registry, then I personally had started patient-reporting registry, which we had to close due to lack of funding, that it's

important to capture the nutrition data, not just the proxy fee was high or low. Why? What did we do? How did we intervene and did we give the right prescription? Did we have the trained personnel to do it -- both physicians and dieticians? Did they know what they were doing when they were prescribing? I think we have enough information now to start packaging it. We should offer -- I would urge the Committee to offer grant funding opportunities for the training we have started, the fellowships we have started to support and really take it to the next level and to help us do curriculums and workshops, which are funded and supported.

It's not on my slides, but after listening to the conversation in which Shawn had asked for the stakeholders, I'm going to make a plea. We would love to see GMDI on the table on this group to bring our expertise, especially when I heard the discussion on GAMT deficiency and the need for our input both in this area and the long-term follow-up registries and all the roles the

dieticians can play. So, I would urge that consideration and with that, next slide, please.

I want to thank everybody for the opportunity and look forward to continued support from the Committee. Thank you very much.

CYNTHIA POWELL: Thank you, Dr. Singh. Those of us who have had the opportunity to work with metabolic dieticians in our practices over the years certainly acknowledge all that you do to make our program successful.

We'll now open it up to questions and comments first from Committee Members followed by organizational representatives. And as always, please state your first and last name and remember to unmute yourself. Use the raise hand feature, if you can.

So, I'd like to ask Dr. Singh, are there formal training programs for metabolic dieticians? In our experience here, it seems that it's often on-the-job training, you know? We recruit excellent dieticians who then, you know, obtain more experience after the begin working

with us.

RANI SINGH: No. There's no formal training program. We have just started the first Academic ECHO training program for twelve weeks using the ECHO model, and actually that's -- the ECHO model is the first in the whole genetics, to my knowledge, no other program has used this yet, and we have done a pilot for twelve-week traineeships and we have had an excellent -- ECHO stands for Extended Community Health Outcomes, most of you probably know, was started by a hepatologist in New Mexico. You have to be licensed and it's a train the trainer program and we just did a pilot, and it was a huge success through SERN, and I would -- and, you know, there have been metabolic universities at the private level, but I do think we need to -- the idea has emerged and the support would be needed. So, no, not at this time other than this ECHO program we have just started.

CYNTHIA POWELL: Thank you.

Michael Warren.

MICHAEL WARREN: Thank you all for those presentations. I was struck by something Dr. Shone said about being able to sort of take the time away from the whirlwind of the current to think strategically and I -- I certainly can say we face that same thing at the Bureau. And so, I just wanted to give a reminder and thanks in advance that one of the opportunities that this Committee has is to make recommendations to us on programmatic operations, and so we welcome those. A number of those have been shared today, and we've certainly been taking note.

One of the things we talk about with our team is thinking, you know, not what can we do in this year and next, which is important, but where are things going to be in five, seven, ten years, and how do we need to be planning now, because as you all know, interacting with federal agencies, the wheels don't always turn quickly. And so, it takes some time, particularly, if we're thinking about changing program design, thinking about funding. So, just to put out that we are

always open and really appreciate your feedback and guidance onto how our programs can meet the evolving needs and particularly how we can anticipate what's coming and what we need to be doing now to prepare for that.

CYNTHIA POWELL: Would any of the speakers like to comment on that or say anything?

SCOTT SHONE: I'll just say, so, Scott Shone, so thank you, Dr. Warren. I mean, I think that your team -- the team who works with this group is amazing. So, Joan and Alisha, and Debi and Mia, are always offering up and, you know, asking and soliciting advice and feedback. So, I agree and I've never -- I don't think anybody would disagree I've ever shied away from sharing my thoughts with them, so I'm happy to continue to do so.

RANI SINGH: I would echo the same. I think it's an amazing experience some of the things we have actually been able to accomplish as a very small group. Many, many volunteer hours by the team, but because of the HRSA funding, we have

been able to do some of these projects -- demo projects. So, we are very thankful for that.

MARCIA FORT: This is Marcia, and I would echo the same thing. Our collaborations with your HRSA team and Bureau team have been amazing and continue to support us and allow us open dialogue and really opened the avenues for improvement and thinking in a forward manner. So, thank you for that.

CYNTHIA POWELL: Any questions or comments from our organizational representatives?

Shawn McCandless, I see your hand up.

SHAWN MCCANDLESS: Thank you. Thanks to each of you. It's really -- this was a fascinating discussion, and it's just a little bit disturbing that there's so much -- so many of the problems -- there's -- there still appears to be a lot of siloization in this area of newborn screening and each person who spoke today, as I was jotting down notes, I was reflecting on the fact that the problems are really universal that each group is facing sort of with the workforce

shortages, we all have the same problems getting people into the field with underpaid and -- people being underpaid and overworked. I suspect we're not along in feeling that way.

I guess the question that I have is with all of these various moving parts in the newborn screening program including our recognition that long-term follow-up is really important but no -- very little standardized approach to long-term follow-up and very little infrastructure to support long-term follow-up, I just wonder what -- what is the best way forward, if any of the speakers have thoughts about what is the best way forward to start to have -- to speak more with a single voice and to have a unified -- a unified infrastructure for driving forward initiatives in newborn screening?

RANI SINGH: I can -- I'm happy to say a few words about that. I -- I just feel that given the technology now, having worked in Public Health Informatics Institute and done a project at a state level, I think that really continuing to

think and figure out ways with technology and looking at systems and working with our state partners, some recommendations for the -- along with the screening for the [inaudible - audio cut out], you know, we talk about registries. We started talking about long-term follow-up components to come to some kind of partnership with the state on how we are going to fund that and to find a home for that so that we can all -- I think everybody's willing to share the data and the informatics is there. How can we identify a home for that.

CYNTHIA POWELL: Melissa Parisi.

MELISSA PARISI: Thank you. This is Melissa Parisi from NICHD, NIH, and I just want to thank all of the presenters for some really provocative and informative presentations.

I think, you know, my question is probably directed more towards Dr. Allen, and the issues that impact subspecialists, particularly pediatric subspecialists, but I think others as well, in that there's always been a bias for

improved compensation for procedural specialists and that has been an issue for a very long while and it seems like the differential is only getting worse rather than better. I'm just wondering, you know, you mentioned some efforts on the part of the AMSPDC -- I'm not sure how you say that acronym -- Workforce Initiative to try to change that paradigm and maybe try to equalize the compensation so that nonprocedural medical specialties can get reasonable compensation for the care that they provide, which is at least as time consuming, if not more, than those that are more procedurally focused, and if there's any thoughts or movement in that direction that you see as positive developments that might help with these workforce issues, because even -- even getting care extenders, I mean, we still have the same issues with those individuals who want them to be trained and specialized, but they're not getting compensated either. So, anyway, I just thought I'd throw that out there for additional consideration on your part.

DAVID ALLEN: Yeah. Thank you for the question and, you know, what I find frustrating about the whole issue of money is, is it -- it's a relative concept, right? It's not like people don't make a reasonable living as a pediatric endocrinologist. Obviously, we do, you know, it's just that there's so much difference between when these medical students are looking at the future, there's just like so much difference between it. To me, it's frustrating that, you know, that they're dissuaded that way.

I will say that that group, the AMSPDC group is how that you say that acronym. I mean, I think they're appropriately looking at both ends of the spectrum. So, one is, you know, this burden of debt, you know, which is almost now inevitable for medical students, you know, either finding new ways of reducing tuition, which would be even better so people don't have to accumulate so much debt or, you know, have targeted loan forgiveness so that they can count on, you know, favorable financial situation after they're done.

I think you have to look at it from both -- from both standpoints.

But what we find fascinating is the first time that those data came out was in 2010, and you could almost track right to then the beginning of the divergence -- the big divergence in the nonprocedural specialties. It's like these medical students, they know what's going on. They know the landscape out there, and they're thinking about it. And so, you know, I think these kinds of data are very difficult to fend off and I think we just have to develop some programs that, you know, they can be educated about to reassure them that they're not making bad decisions. I think that's what they fear is that they're making some kind of a bad professional decision to go into these fields.

But I am optimistic. As I mentioned in my presentation, you know, this -- in this January, you know, there was this acceptance by the payers of what's called time -- time determined coding, you know, which has been a

major -- major significance, I think, certainly for pediatric endocrinology to -- as a way of increasing our VU production. It's a small step, but I think it's a step in the right direction and the more accountable care takes over to look at the global management of patients rather than just encounter-based management, I think that will -- that should help us in the future.

CYNTHIA POWELL: Jed Miller.

JED MILLER: Yes, hi. Jed Miller, Association of Maternal and Child Health Programs. First of all, I've been enjoying the pronunciation of the acronyms here during this presentation. It's been really good stuff.

My question actually is for Dr. Shone. I'm curious about your thoughts about the interface specifically between the lab and the follow-up side, and I know that, you know, the training, there's no dedicated training, but I'm curious if you have any thoughts about if there were to be training, would -- would dedicated training on that front -- the actual interface

itself -- be of value? And on a related, I guess, note, are you aware of any state programs where there are liaisons between the, you know, the lab and the follow-up side that look at process -- broader process-type considerations, and wondering about any comments on that.

SCOTT SHONE: Sure. So, I think there are certainly examples of how elegantly lab and follow-up work together. I think that when -- when we don't, it is a great detriment and risk to the newborns whose health we are trying to protect here. And so, at every level -- and I think there is -- there are a plethora of examples, and I want to be cognizant of the time, Jed, so I'm happy to chat offline too or just follow-up with notes perhaps as part of the record of the meeting. But in terms of models of lab and follow-up interactions where they are co-located physically and organizationally, they are dislocated organizationally and physically, and then some where you have contract labs but the follow-up remains in the state, and that's just three of the

few that are, you know, I think the top three examples that we have and -- and they all function well. We've had presentations on all of those -- on all those fronts at this Committee over time. I do feel that the communication and having been through this through, you know, organizational site visits from our groups, whether it's New Steps or otherwise, to help identify opportunities for better communication, information technology uses, to better streamline a lot of that is there and I think we all have room for improvement along the way.

In terms of I don't know if your question also touched on training, I don't know that there's a training lean on the coordination between lab and follow-up. I'll have to think a little bit more about that one. But I do feel that we've seen great examples of how the coordination works. I think what we just need to realize always is that it's bidirectional, right? So, it's not simply the lab test sends a result and we're done with it. Like the most successful

programs are those that have this feedback mechanism so we understand what's going on. A lab operating in a silo cannot improve, cannot undergo quality enhancements, and a follow-up program that doesn't understand what's happening at the lab doesn't, you know, doesn't benefit from -- from why things are the way they are, and I think the best ones we've seen are the ones who have those routine communications.

So, it's a long way of just saying I have more thoughts on that later.

JED MILLER: Thank you.

CYNTHIA POWELL: We are running short on time, but I want to give those who have had their hands raised the chance. Max Muenke.

MAXIMILLIAN MUENKE: Thank you. I think my hand blends in with my bookshelves. I just wanted to say I was reminded of the fact that Dr. Berry is here. Thirty-five years ago, Dr. Berry was my attending when I was in biochemical genetics training in July of 1986 at Children's Hospital in Philadelphia. This session was a

fascinating session. I find that it speaks to the fact that we have to work closer together and even though Dr. Singh and I, we are, courtesy of HRSA and the National Coordinating Center and Regional Networks, we are on similar cause and I would want to have a closer collaboration between the American College of Medical Genetics and Genomics and SIMD and the Society that Dr. Singh is part of because in the end, it is still the way, as you point out, Cindy or Dr. Powell, that it's sort of we are all siloed and that the training happens on the job, and I find we need this training for metabolic genetics and metabolic dieticians. We need that more around the country. We need that in different institutions, and to me, if we don't collaborate, we won't get there. So, to me, it's just a plea for collaborations and I wanted to reach out here to Dr. Singh and we fill follow up on another call just to -- to just point out a year ago my predecessor, Dr. Mike Watson, initiated under HRSA's guidance initiated the -- the Workforce Study and that was mainly for

clinical and laboratory geneticists, but it did not include dieticians. It did not include other health care professionals who work in the same field, and I would very much want to invite all of us to be part of a -- of a future workforce study that we're planning now, planning in the 2021, 2022, or 2023 that will involve all of us here from dieticians to genetic counselors to PAs. There's a society for genetic PAs and then obviously SIMD and then genetics and laboratory genetics. So, a little wide, but thank you for this session. This was an amazing session this afternoon and today. So, thank you.

CYNTHIA POWELL: Susan Tanksley.

SUSAN TANKSLEY: Thank you. Susan Tanksley, Association of Public Health Labs. I'll just make a few comments, going back to Scott's talk, but really, I think it kind of fits for all the talks, you know, we've had. I've been co-chairing the APHL Workforce Work Group for a while now, and we've really struggled with trying to figure out how -- how do we make an impact and we

started out with the just, you know, how do we -- how do we get people interested in public health labs, or in this case, in any of these professions, and then the engagement piece, once you get them in, how do you not just be a training ground, you know, for their next job and how do you keep them interested in what they're doing so that they want to continue with you and grow with you. And more specifically to the newborn screening workforce piece and trying to figure out, what is it that we need in a newborn screening program, whether it be the laboratory or the follow-up piece, and the fear is that if we establish a minimal staffing guideline that somebody is going to take that and go, well, that's all you need then, you know? So, we don't want to do more harm than good with whatever we come up with. So, we've been really wracking our brains trying to figure out how to approach that and not just what's -- what's minimal staffing, but what are really the minimal things that newborn -- what are -- what are all the things

that newborn screening programs at minimum should be doing so that it includes the quality improvement piece, so that it includes the ability to grow and to make yourself better, not just get by?

Thank you so much to all the speakers today. It was really engaging and interesting.

CYNTHIA POWELL: Gerry Berry, we'll let you have the last word.

GERARD BERRY: Oh, thank you. Gerry Berry, SIMD. Thanks, Max. You know, to have newborn screening be effective, you really need a whole team of individuals all working together. I just want to support what Dr. Rani Singh said. She is so true -- she's so right about the comments that she made. Without -- without newborn screening follow-up with dieticians plan to complete an important role, we just -- we just won't have an effective system. They're so important. We've learned so much from them over the years and I -- I would like -- would support their involvement as much as possible. They're

absolutely wonderful in making newborn screening work. Thank you.

CYNTHIA POWELL: Thank you. Yes, I think consideration again for including their organization in our -- among our organizational representatives.

So, I'd like to thank all of our speakers for this afternoon. They were excellent presentations. You really all hit the mark and certainly engaged the group, who are very interested in this. So, I'm thinking that in follow-up, you know, things like a white paper, publication, if anyone is interested in working on something like that, please let us know. Let me know, let Mia know.

NEW BUSINESS

So, finally for today, do Committee Members have any new business or announcements? Let's see, Scott Shone.

SCOTT SHONE: I would just like to say given that we are halfway through August, I want to wish everybody in the Newborn Screening

Committee for September Happy Newborn Screening Awareness Month. You'll notice Natasha Bonhomme's background celebrates Newborn Screening Awareness Month. So, let's celebrate that then also to my colleagues in the Public Health Lab, September also is National Public Health Laboratory -- goodness gracious, I just blanked -- Public Health Laboratory Awareness Month. And so, public health lab and newborn screening, basically everything I talked about in my talk this afternoon, I totally forgot to say it while I was speaking earlier. So, thank you, Dr. Powell.

CYNTHIA POWELL: Thank you. Our next meeting will be in November -- November 9th and 10th.

Oh, Annamarie. Sorry, I didn't see your hand.

ANNAMARIE SAARINEN: Oh, it was truly not important until Dr. Shone said that. So, just Annamarie Saarinen. I was just going to say it's also the 10th anniversary of the addition of CCHD screening to the panel, which I know is an unusual

one that had lots of lessons learned and continues to have lessons learned from it anyway. But just from my own advocacy, I know we're really sort of acknowledging and celebrating that milestone.

CYNTHIA POWELL: Great, thank you. Very important.

All right. Well, with that, I will adjourn the meeting. Thank you all for your participation, and I'll look forward to getting back together. I believe it's going to be virtual again, unfortunately, but in November. Take care, everyone. Bye bye.

[Whereupon the meeting was adjourned at 2:00 p.m.]