The Advisory Committee on Heritable Disorders in Newborns and Children

U.S. Department of Health and Human Services

Virtual Meeting

Day 2

Friday, May 14

10:00 AM

Committee Members in Attendance

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

Kyle Brothers, MD, PhD

Endowed Chair of Pediatric Clinical and

Translational Research

Associate Professor of Pediatrics University

of Louisville School of Medicine

Jane M. DeLuca, PhD, RN

Associate Professor

Clemson University School of Nursing

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 in Attendance

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

Ex-Officio Members in Attendance - continued

National Institute of Health

Melissa Parisi

Designated Federal Official in Attendance

Mia Morrison, MPH

Genetic Services Branch

Maternal and Child Health Bureau

Health Resources and Services Administration

Organizational Representatives in Attendance

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

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

Organizational Representatives in Attendance - continued

Genetic Alliance

Natasha F. Bonhomme

Vice President of Strategic Development

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 AND ROLL CALL

Cynthia Powell:

Good morning, everyone. Welcome to the second day of the May 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. Kamila Mistry?

Kamila Mistry:

Here.

Cynthia Powell:

Mei Baker?

Mei Baker:

Here.

Cynthia Powell:

Jeff Brosco?

Jeff Brosco:

Present.

Cynthia Powell:

Kyle Brothers?

Kyle Brothers:

Here.

Cynthia Powell:

Jane DeLuca?

Jane DeLuca:

Here.

Cynthia Powell:
Carla Cuthbert?

Carla Cuthbert:
Here.

Cynthia Powell:
Kellie Kelm?

Kellie Kelm:
Here.

Cynthia Powell:
And for HRSA today, Joan Scott?

Joan Scott:
Here.

Cynthia Powell:
Shawn McCandless?

Shawn McCandless:
Here.

Cynthia Powell:
Melissa Parisi?

Melissa Parisi:
Here.

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

Annamarie Saarinen:

All right, here.

Cynthia Powell:
Scott Shone.

Scott Shone:
Here.

Cynthia Powell:
And for our organizational representatives, Robert Ostrander?

Cynthia Powell:
Deborah Friedenberg?

Cynthia Powell:
Maximillian Muenke?

Maximillian Muenke:
I'm here.

Cynthia Powell:
Stephen Ralston?

Cynthia Powell:
Jed Miller?

Jed Miller:
Here.

Cynthia Powell:
Susan Tanksley?

Cynthia Powell:
Chris Kus?

Chris Kus:
Here.

Cynthia Powell:
Shakira Henderson?

Shakira Henderson:
Here. Hope you guys can hear me today.

Cynthia Powell:
Yes. Thank you.

Shakira Henderson:
Awesome. Thank you.

Cynthia Powell:
Jennifer Kwon?

Jennifer Kwon:
Here.

Cynthia Powell:
Jacob Hogue?

Jacob Hogue:
Here.

Cynthia Powell:
Natasha Bonhomme?

Natasha Bonhomme:
Here.

Cynthia Powell:
Siobhan Dolan?

Siobhan Dolan:
Here.

Cynthia Powell:
Cate Walsh Vockley?

Cate Walsh Vockley:

Here.

Cynthia Powell:

Georgianne Arnold.

Georgianne Arnold:

Here.

Cynthia Powell:

Is there anyone whose name I called who wasn't able to respond, but is present? Okay, thank you. I'll next turn it over to Mia.

Cynthia Powell:

Oh, I'm sorry. One thing before we start. Okay. Today we'll begin with an overview of the National Survey of Children's Health data. We have one individual registered to give public comment on updates to the Committee's evidence review process. After the public comment period, we'll break until 11:40 Eastern time. For our final session of the meeting, the Committee will hear a panel presentation on workforce issues related to short- and long-term follow-up. I'll now turn it over to our Designated Federal Official Mia Morrison to provide guidance for participation on the webinar.

Mia Morrison:

Thank you, Dr. Powell. For members of the public, audio will come through your speakers, so make sure that your computer speakers are turned on. If you can't access audio through your computer, you may dial into the meeting using the telephone number in the email with your Zoom link. This meeting will not have an attendee chat feature, but we do have the public comment periods scheduled later today.

Mia Morrison:

Committee Members and Organizational Representatives, audio will also come through your speakers and you will be able to speak using your computer microphone. If you cannot access the audio microphone through your computer, you may need to dial into the meeting using the telephone number in the email with your user specific Zoom link. Please remember to speak clearly, state your first and last name to ensure proper recording for the Committee transcript and minutes. The chair will call on Committee Members first and then Organizational Representatives.

Mia Morrison:

In order to better facilitate the discussion, Committee Members and Organizational Representatives should use the raise hand feature. When you would like to make comments or ask questions, simply click on the participant icon, and choose raise hand. Please note that depending on your device or operating system, the raise hand feature may be in a different location. To troubleshoot, please consult the webinar instructions page in your briefing book. Next slide, please.

Mia Morrison:

To enable closed captioning, please select the closed captioning icon from the Zoom task bar. From the menu that appears, select show subtitles. And I will now turn it back over to Dr. Powell.

OVERVIEW OF THE NATIONAL SURVEY OF CHILDREN'S HEALTH

Cynthia Powell:

Thank you, Mia. Today. I'm very pleased to welcome Dr. Julie Donney and Dr. Reem Ghandour from HRSA's Maternal and Child Health Bureau, Office of Epidemiology and Research, who will provide an overview of the National Survey of Children's Health.

Cynthia Powell:

The National Survey of Children's Health provides rich data on multiple intersecting aspects of children's lives, including physical and mental health, access to quality healthcare, and the child's family, neighborhood, school, and social context. The National Survey of Children's Health is funded and directed by the Health Resources and Services Administration Maternal and Child Health Bureau. This is an opportunity for the Committee to become familiar with the available national survey data, especially data on genetic conditions and how they go about collecting the data and learn about potential future innovations and new approaches for the survey.

Cynthia Powell:

I'll now introduce both of our speakers. Dr. Ghandour is the director of the Division of Epidemiology within the HRSA Maternal and Child Health Bureau's Office of Epidemiology and Research. Her individual portfolio includes conducting original research on a variety of maternal and child health issues with a special focus on children's mental health, early learning, and children with special healthcare needs.

Cynthia Powell:

She has been recognized by the Coalition for Excellence in Maternal Child Health Epidemiology twice, winning the Maternal and Child Health Young Professional Achievement Award in 2011, followed by the Effective Practice Award in 2020. And in 2016, she received the Maternal Child Health Bureau Director's Award at the Title V Federal-State Partnership Meeting. Dr. Donney is part of the National Survey of Children's Health team, where she conducts original research on child and family health. Examples of her previous research include promoting flourishing during early childhood, mental health of teen parents and their children, parenting practices of Latino immigrant parents of

adolescents, and child abuse prevention. Dr. Donney holds a PhD from the University of Maryland School of Public Health and an MPH from the University of Montana's School of Public and Community Health Sciences. I will now turn it over to Dr. Ghandour.

Reem Ghandour:

Thank you so much, Dr. Powell, and thanks to all of you for allowing us to take a little bit of time this morning to talk about the National Survey. Let me just do a quick check right now. Can everybody hear me all right?

Cynthia Powell:

Yes.

Reem Ghandour:

Okay, fantastic. So today, Julie and I are going to do sort of a high-level picture of the survey just to familiarize you with the survey as a resource, and we will be providing some key estimates just to also give you a snapshot of some of the data that are available to you. And then ultimately, we're going to wrap up with just giving you a sense of where the survey is going. We're happy to keep this informal, so if you have questions, please feel free to use the raise hand function and I'll do my best to sort of keep an eye on that as well as we go.

Reem Ghandour:

Next slide please. So, our goal today is really four-fold. I will provide an overview of the survey and just touch on a few key differences. I think many people know that the survey was redesigned dramatically in 2016, but there's still a fair number of folks who are used to working with the previous National Survey of Children with Special Healthcare Needs. So, I'll just make sure that we're all clear about kind of where the survey is now and how it's administered. Julie is going

to present some key estimates using a combined data file from our 2018-2019 survey. I will talk about how you get to the data if you want to start using it yourself, and then we'll give you a sneak peek into sort of what's coming in the next year or two with the survey.

Reem Ghandour:

Next line. And next slide. Thank you. So just as was briefly mentioned, the National Survey of Children's Health is sponsored, it's funded and directed by HRSA's Maternal and Child Health Bureau. But I think it's really important to note that we have co-sponsorships from other parts of the federal government, particularly CDC. We work quite closely with a couple of different divisions there, as well as USDA. We are now conducted by the U.S. Census Bureau. So, for folks who were familiar with our previous surveys, we used to be conducted as a telephone survey through the National Center for Health Statistics. This is now an address-based survey that's conducted using the master address file from the U.S. Census Bureau. One of the other ways the survey is dramatically different now is that it is an annual survey. So, it remains cross-sectional, but it is an address-based survey. And about 75% of our data are collected via the web with the remainder being collected via paper and pencil questionnaires.

Reem Ghandour:

Although the administration and the mode of the survey is dramatically different than what we did in the past, the survey remains true to our overall goal, and that is to collect information on health and wellbeing of children from zero to 17, as well as the sort of spectrum of community, family, healthcare related factors that can influence children's health. And we still provide national- and state-level estimates, so that remains important from a policy perspective. What we do is the survey is in the field for approximately

nine months. We go from June to roughly January, and then we release the data the following October. So for those of you used to federal data collection, this is almost lightning speed, not quite real-time, but pretty darn close. And so you can usually look for the latest data release sometime the first Monday in October. Next slide.

Reem Ghandour:

We cover eight broad content areas, universally for all age groups. We do have separate surveys, slightly different for zero- to five-year-olds, six- to 11-year-olds, and 12- to 17-year-olds. But what's presented here is really the core content that runs across the survey. Where I think this Committee may be most interested, is in general health where we have our activity, or I'm sorry, our condition list, which includes blood disorders as well as other genetic conditions. And it also gets at activity limitations and some of the functional limitations that may be relevant to some of the work that you're doing. And then in addition to infant health, pre chambers, low birth weight for the zero to five-year-olds, we cover infant feeding and infant sleep position. We have a range of content that gets at both the access to and utilization of health care services, particularly specialists in addition to the normal preventive care, as well as the quality of the care that is received, and specifically looking at the receipt of care in a medical home.

Reem Ghandour:

We go deeper than any other survey with regard to health insurance. So, we're looking at not just the status and the types, or are you insured and what's the source of that coverage. But we do ask parents a series of questions about adequacy. So, folks are able to report on whether or not their child's needs are actually being met. And then we have a range of items

that were brought over from our previous National Survey of Children with Special Healthcare Needs that focus on the impacts within a family of caring for a child with a special healthcare need without a genetic condition or some other condition. And then of course we have content that's related to family and neighborhood factors. For very young children, we've also now included a range of content related to healthy and ready to learn, which is specifically, sort of school readiness, in the simplest terms. And then for older children, we include content around health care, transition planning. Next slide, please.

Reem Ghandour:

One of the biggest drivers of our content is what our Title V and our state partners who work within the title universe need. And we are the source for 20 of the Title V performance and outcome measures. Some of them are bolded here, as they may be of particular interest to this work group. We're also the data source for 15 Healthy People objectives. We remain a foundation for much of the work that the CDC does in their National Center for Birth Defects and Developmental Disabilities. And we are a benchmark for our home visiting and healthy start programs and some of their performance measures. And others, we retain the CSHCN Screener, and so we're the primary data source for Children with Special Healthcare Needs, prevalence and impact. Next slide, please.

Reem Ghandour:

Just to give you a sense of the utility of the survey. These are just a snapshot of a couple of the ways that the survey data get used. Sometimes we're very intimately involved in this kind of work. So, we are the data source for the state estimates that RWJF puts out every year on childhood obesity. And we usually run those data with them and help them make decisions about what should be presented. In other cases, we find out

with the rest of the world. So, when Starbucks used our family impact data to make changes to their personnel policy, we found out with the rest of the world on CNN. But we will take any use of the data. And we're grateful that both the public and private sector are able to use these data to improve both our understanding of the health status of kids, but also their needs. Next slide please.

Reem Ghandour:

So that's just a really high overview of the survey. I want to turn it over to Julie, to provide a little bit more in depth on the health conditions that are in there and some of the other indicators that we have.

Julie Donney:

Awesome. Can everybody hear me okay? Okay. So, this slide gives you a sense of where genetic inherited conditions fall among the overall prevalence of current and lifelong health conditions among children. So, in 2018 and 2019, allergies were the most commonly reported health condition among children at 19.6% followed by ADD and ADHD at 8.7, and anxiety at 8.5. So other genetic and inherited conditions fall at number nine at 3.8%. Next slide.

Julie Donney:

Taking a closer look at heritable conditions, this graph compares 2018-2019 estimates with 2016 and 2017 estimates in the blue. At the top, other genetic conditions increase from 3.02% to 3.78%, and that was a statistically significant increase, but there were no statistically significant differences in the percentages of children with blood disorders, down syndrome, or cystic fibrosis. Next slide, please.

Julie Donney:

In 2018, questions about newborn screening were added to the blood disorder and other genetic inherited

condition questions. You can see them they're outlined in the red, were any of these blood disorders identified through a blood test done shortly after birth. These tests are sometimes called a newborn screening. Next slide.

Julie Donney:

And these pie charts show the percent of children with blood disorders and inherited conditions who were identified by newborn screening. So, the blue sections represent the percentage of children who were identified by newborn screening, and the red sections represent those not identified by newborn screening, so a different way. So, among children with genetic and inherited conditions on the left, 20.2% were identified by newborn screening, and among children with blood disorders on the right 66.4% were identified by newborn screening. Next slide.

Julie Donney:

So, this graph shows the differences by household income, insurance type, education, and maternal age. The blue bars represent children with genetic disorders who are identified by newborn screening, and the red represents the children with blood disorders who are identified by newborn screening. The percent of children identified by newborn screening decreased as income increased, and then more children with public insurance were identified by newborn screening than children with private insurance. There were no major significant differences based on education or maternal age. I'll just note here that the numbers for maternal age were quite small, even though it looks as though the percentage of children identified by newborn screening were much higher among women who were 42 and above, those numbers were very small, so they are unreliable. Next slide, please.

Julie Donney:

These graphs show the percent of children with blood disorders or inherited conditions who have zero, one or two functional limitations. So, the National Survey of Children's Health captures 12 functional limitations. These are difficulty breathing, eating and swallowing, digesting food, using hands, moving around, difficulty concentrating, walking upstairs, dressing, doing errands, or if they have chronic physical pain, deafness or problem hearing, or blindness or problem seeing. So, on the pie graphs, the number of functional limitations increase as the color gets darker. So, for blood disorders on the left, a majority of children, 53.7% had no functional limitations, 19% had one functional limitation, and 27.3 had two or more. Among children with genetic conditions, 35.3 had no functional limitations, 26% had one, and 38.5 had two or more. I also wanted to note on this slide that other heritable conditions are not shown just because the numbers are so small, and so the estimates are unreliable. Next slide.

Julie Donney:

This slide shows the percentage of children with each heritable disorder who had unmet needs there in the blue, or whose parents experienced frustration, getting care in the red. The black and yellow dotted lines show the percentage among all children to kind of give you an idea of what that looks like. Among children with heritable conditions, children with cystic fibrosis had the most unmet needs for healthcare and more of their parents experienced frustration getting care. Next slide.

Julie Donney:

And finally, this last slide that I'll show you shows the number of hours per week that parents spent either arranging health care for their child or providing health care at home. The darkest blue represents reports of children not needing either care arranged or

provided, and then the light blue is less than five hours, and the red is five or more hours. And there's a lot of variation based on the specific health condition. Parents reported the most time spending, arranging, or providing care at home among children with cystic fibrosis. 32.4% reported five or more hours per week arranging care, and 53.4... Sorry, 54.3 reported five or more hours providing care at the home.

Julie Donney:

Among the heritable conditions, parents of children with blood disorders reported the least amount of time, arranging care. A majority of those children did not need or need, sorry. The majority of those children did not need arranging healthcare or providing health care in home. Next slide, I'll turn it back over to Reem.

Reem Ghandour:

All right, thanks, Julie. I think what was helpful about those data points is that it really just gives you a snapshot of kind of how you're able to stratify and look at sort of populations of interest and kind of their health status as well as their needs. Next slide, please.

Reem Ghandour:

So, the next couple of slides are just designed to give you a sense of where you go, if you want to get to the data, depending on your level of comfort. So, this is the HRSA web page. It really is a one-stop shop to get to pretty much anything, either that we've done as internally as kind of a data brief or what have you, but also can get you to all of the technical documentation as well as the data sets. Next slide.

Reem Ghandour:

This is the data resource center. We fund this out of Johns Hopkins with Dr. Christie Bethell, and what's helpful about this particular website, if you're not

familiar with it, it's a great kind of point and click. If you just want to get the quick prevalence of any condition of the 25 or so that we ask about, then you can do that. You can look at national estimate, state estimates, and then there's also an opportunity to do some very simple stratifiers by race, ethnicity, poverty, et cetera. Next slide.

Reem Ghandour:

For those of you who are more interested in actually diving into the data, you can download all of the micro files from the Census Bureau. At this point, we're not releasing combined files, which is probably of particular importance to folks who want to look at what disorders are genetic conditions that are relatively rare compared to some of the other conditions that we have on the survey. So, you would likely need to combine multiple years. You can certainly that, and we have technical documentation about how you would handle the waiting and how you upend the files, et cetera, if you need that assistance. Next slide.

Reem Ghandour:

And I'm just going to wrap up now with just looking at a couple of the different ways that we are innovating with the survey and some of the new content that you can expect. So here is just a snapshot. It should say, sorry, the slide should say 2020 and beyond, but we have a little bit of new content for 2020. We actually tried to keep it relatively tight. These data will be available this coming October. We're going to be starting to internally look at the data files shortly here. And it is our first year where we had state over samples. And I'll have a slide on that in just a moment. In 2021, which will go out this June, we have new content that's sponsored on early childhood nutrition. We have some five questions related to COVID-19 impacts, and we've added an ACEs item, an

adverse childhood experiences item, that focuses on discrimination due to physical ability.

Reem Ghandour:

This is analogous to our race and ethnic discrimination item that's already on there, and we'll be doing a national oversample of young children. So, this is coming up. The 2021 sample is likely to be very robust. Even the 2020 is actually has about 41,000 observations. So, we are really building up a sort of a spectrum of sample that you can work with depending on what your research area of interest is. For 2022, we will be revising and finalizing, God willing, our content around early childhood learning. We're adding housing insecurity, as well as some content around disordered eating, parental leave, and urgent care. Next slide.

Reem Ghandour:

Next slide, maybe. I don't know what's happening. There we go. Just a quick note on state over sampling. In 2020, we offered our state partners the opportunity to purchase over samples. And some folks did this because they just wanted to be able to do more robust analyses of smaller population groups. And others, and this is really, I think, more relevant for more than less of the states. They wanted to be able to do sub-state analyses. And so, since this works with each state independently to come up with a plan that meets both their budget and their analytic needs, and it costs anywhere from between 20,000 to 500,000, depending on the state and the analytic area of interest. So, we are planning, or had a total of nine in 2020, and are sort of maintaining some and swapping some out as we go forward in time. Next slide, please.

Reem Ghandour:

And this is just a quick map of where we were and where we think we are headed. Just depends on Pennsylvania and New York, going forward. Next slide.

Reem Ghandour:

This is the national over sample that we'll have of young children. So, this is going to be great for folks who really want to dig into not just some of the early learning, but also kind of early childhood behaviors and family behaviors that promote early learning. We have five new questions that get at consumption of fruits, vegetables, sugar-sweetened beverages, as well as time spent playing outdoors. And so, this is one of those opportunities where the national over sample of young children is really raising all boats. So, we will have a much more robust sample overall for the 2021 survey. Next slide. And this is just my contact information. You should feel free to shoot me an email, and Julie and I can triage as needed. But again, this is just designed to give you a snapshot. We're happy to take any questions you might have.

Cynthia Powell:

Thank you both very much. It's really informative. Interesting to find out more about the national survey. We do have some time for questions and comments. We'll start with Committee members first, and then we'll go to organizational representatives. Please remember to state your name before speaking, and we'll start with Jeff Brosco.

Jeff Brosco:

Thank you. This is Jeff Brosco, and I want to start by thanking you, Reem and Julie, and all your team. This is a national treasure. The ability to get this information and use it is just essential for all the work we do. I wish we could oversample in every state, it's sometimes tricky for our states to figure out a way to pay for oversampling. It's frustrating.

Jeff Brosco:

I did have a question for you, Julie, in particular about the slide that looked at newborn screening diagnoses, that looked at level of poverty. And in particular, I wonder if you've had a chance to look at this because on one hand it could be that certain conditions are more prevalent in certain populations, but it could also be evidence that newborn screening helps reduce inequity in health care. So, I wonder if anyone's really looked at that more closely and if you have any ideas about it.

Julie Donney:

You know, I'm not sure if anybody's looked at it more closely, I'll ask Reem, but I did find that really interesting, and I was hoping that you all might be able to explain that a little bit more. I was wondering, the question that came to my mind was, is this saying that, if these conditions are not caught with newborn screening that they go undiagnosed as they get older or I don't know. I was looking forward to learning more from you.

Jeff Brosco:

So, this is Jeff Brosco again. We actually did a paper looking at SCID some years ago. It showed before newborn screening. We thought SCID was primarily in non-Hispanic whites. And then after a couple of years of newborn screening in California, it turned out it was quite the opposite -- that it was Blacks, Hispanics, and Asians who had a much higher level of SCID in the prevalence. And so, we think that this is probably about the universal nature of newborn screening and shows how newborn screening is one of the ways that we improve equity in our healthcare system. But it could also be that these are confounded by who more is more likely to have a condition, and so I think it's worth digging into more.

Reem Ghandour:

To my knowledge no one has jumped in on that.

Reem Ghandour:

You know, I think this is one of the areas, where we haven't seen a ton of work done with it. So we'd love to see more, especially as the sample gets bigger and there's more to work with. We'd love to see more work done in that area.

Cynthia Powell:

Melissa Parisi?

Melissa Parisi:

Thank you, this is Melissa Parisi from NIH. So, I wanted to thank you as well for this important work. And I remember when you were talking about adding the newborn screening questions. My question for you is that obviously, as you expand the survey and you want to be adding more questions, at some point, you must reach a saturation level where you can't ask everything you want to ask. So, what's the process for actually determining when you may retire questions or rotate them off of the survey.

Melissa Parisi:

And then I guess I also had a follow-on question about the state oversampling, and some of the reasons for that. I saw that you mentioned race and ethnicity targets, but I'm wondering if there were other reasons why states choose to oversample and how that process works a little bit better.

Reem Ghandour:

Sure. I can take both of those and there's lots more to say. But in terms of content decisions, you're absolutely right. We're actually very sensitive to the length of time that it takes to get through the survey. We didn't want to be in the same position that NHIS was

in, where it was at getting at 60 minutes and they were really losing folks, even as an in-person survey. So, we're at 37 minutes right now on the web survey. And we know in particular that the instrument for children that are zero to five is the longest and that is because of everyone's interest in life course and early intervention, that is also the place where everybody wants to add content. So, we have a pretty robust process, where, folks, it takes about two years to be really honest. If somebody wants to propose content to really evaluate and actually kind of make it through the gauntlet and get added, that takes about two years.

Reem Ghandour:

So, we ask folks first to propose, preferably validated questions, usually not more than one to three, sometimes five, you see places where people have been able to sponsor up to five. We do a cognitive testing with census every single year of the survey. So, we do really try to make sure that everything that is on there works well with a range of respondents. And then we take a really hard look at the Bureau's priorities and populations that we are charged with serving legislatively, as well as the field. And we try to make the best educated decision we can based on the pros and cons.

Reem Ghandour:

To date, we have very little content that has come off. What we've done is we worked closely with census to try to find efficiencies within the survey design to make it less burdensome. But the truth is we are getting to that place where we're going to have to make some hard decisions. And if content has simply not been used in any sort of papers that we can find, if it's really not a priority area for the Bureau or close partners, then that's sometimes a place where content would fall off. Especially, if it's not necessarily content that we feel like is the best way to get at a particular

question, or if parents aren't necessarily the very best reporters.

Reem Ghandour:

So, I'd like to say, it's a nice linear process. It's not, it's very iterative. And we rely a great deal on our partners in the Bureau, Joan and her team in particular kind of really help us and others in her position, sort of really help us make that determination and make some of the tough calls.

Reem Ghandour:

To your question about oversamples. We've had states who wanted to really look at children with special healthcare needs in particular, although we do our best, and we do have an over sample of kids with special health care needs, I think when you really want to drill into that population on a somewhat more granular level. You still need a more robust sample. So, some states have looked at CSHCN status. Some have been interested, in particular, racial and ethnic groups, and some have been interested in a very particular area of content. They have a governor who really cares about early learning or early childhood, and that's where they want to put their money.

Reem Ghandour:

I will say this, I think Jeff, one of your points about wanting to give more states the opportunity to purchase oversamples. One of the things we're trying this year with Pennsylvania, is that we're allowing them to pull it from their block grant allocation. Which for many states, the block grant is... What an oversample cost is, frankly, pencil dust on their block grant, but it's a more streamlined opportunity for them if they want to allocate those funds, and census and our team will work with them to create a plan that is within their budget.

Reem Ghandour:

Hopefully, I answered all of your questions, at least marginally.

Cynthia Powell:

All right, due to our time limitations, Mei Baker, Natasha, Jacob, and then we'll have to go onto our next session. Mei Baker?

Mei Baker:

Sorry. Okay. Mei Baker, a Committee Member. My question is more for clarification. And when we use a newborn screening, I'm more curious because Julie... defined is that you identify to detect it at a birth. I'm not so... I just want to be clear because for this group, when we are told we have a newborn screening we have a panel in mind. And just based on the data you presented, for example, you talk about the blood disorders. I see the slides, you have a sickle, and you got thalassemia and also you put a hemophilia in that. So it's... Why you look at that? Identify from where? Because I know hemophilia is not in a newborn screening panel. So could you just clarify, when do you use a newborn screening, if I understand correctly, it doesn't mean the newborn screening panel we address.

Julie Donney:

Maybe we could show the slide again with the newborn screening question.

Mei Baker:

Yeah. When you get the slides up. I hope I asked clearly, because you said that you have inherited disorder above a 20 also. So, to me, do the math. So, I thought maybe when you talk a newborn screening as a conception, or really indicated these, disorders, several disorders, so you identified at birth can be done through state newborn screening program also could be done at bedside, like in a hospital. I just want to know if I understand correctly.

Julie Donney:

Yeah. I actually I'm not sure. And Reem, I'm not sure if this was part of this discussion. I know that it says the tests are sometimes called newborn screening...screening. And I don't know if that would, account for that, but sometimes it's maybe not called newborn screening, but I don't know if you have anything to...

Reem Ghandour:

Yeah. So, the only thing, so Mei, I think you're driving on an important point, which is the level of specificity that we're getting at when we ask some of these questions. And one of the sort of perennial challenges we have is that this is a parent reported survey and it's self-administered, meaning we don't have the advantage of an interviewer who can sort of ask probing questions or get in any more depth.

Reem Ghandour:

So, the questions you see here are basically the wording that we were able to come up with, working with Joan and her team, and others, I believe who are on this call, as well as after we had the opportunity to do cognitive testing. And we made sure that parents could actually understand what we were asking. So, to your point, it's possible that there is variation in what is being reported. And at least in the moment, when we did this, it felt like the best we could get, but we may be missing or mis-characterizing some of the experience of testing.

Mei Baker:

Thank you.

Cynthia Powell:

Natasha Bonhomme?

Natasha Bonhomme:

Hi, Natasha Bonhomme, thank you. This is great. It's really wonderful to see these questions added in. I have, I guess, a comment and two questions. One, I think it's really interesting to see the data from the cystic fibrosis parents. Considering the fact that some would say that's an area that has had a lot of research and a lot of resources and so much done from a clinical perspective, but also the advocacy perspective. So that's just something that could be interesting to try to unpack.

Natasha Bonhomme:

My two questions, one's a technical one. For the other conditions, when you go into the database, are you able to see all of what was kind of written in, in terms of other conditions, is that available? Or, is that all collapsed under other genetic conditions?

Reem Ghandour:

So for that write-in, you would have to go in to one of the RDCs, it's not on the public use file, so you'd have to write a request and get in there.

Reem Ghandour:

I have not, just from a resource perspective, our team, we're actually a pretty small team of about five people running the whole survey. So we have not had a chance to dig into the write-ins, but I think that's obviously an option and something that you could propose to do.

Natasha Bonhomme:

Great. And then lastly, do you ask any questions about prenatal diagnosis? I think at a time now, when there are, there's a bit of an overlap between prenatal diagnosis and what you can be diagnosed with your newborn screening and early childhood, just seeing if there's any information there? Even though...

Reem Ghandour:

That's a really interesting question, and one that actually just amongst our team, many of us are parenting young children and have been pregnant between the time that the 2016 survey was written and then even the 2020 questionnaire. And they uptick of like early prenatal screening is really different. Even in that time period, we've been debating whether or not we should add content on that. We weren't quite sure what the content should be yet. So, it's on our radar. We just haven't figured out exactly how to do it. And if you have thoughts, we would love to hear it. So, we're always open to that.

Natasha Bonhomme:

Absolutely. Great. Thank you so much.

Cynthia Powell:

Jacob Hogue?

Jacob Hogue:

Yeah. I think more of a consideration on top of what you were just speaking of as well as about Jeff's question. I wonder how much the general accessibility of prenatal diagnosis and preconception testing and screening is influencing that difference that you're seeing based on income level. Where I think, fortunately, newborn screening is a universally available testing modality, but there's certainly differences in access to care for prenatal testing. So those families that feel that the condition was identified based on newborn screening is probably different in populations that knew they were carriers for those disorders based on having the accessibility to that testing previously. So, I wonder how much that's influencing that difference, rather than being a difference in the rates of those disorders in those populations. Thank you.

Cynthia Powell:

Chris Kus.

Christopher Kus:

Yeah, you had mentioned in the 2021 survey, you're going to have six questions about COVID-19 related impact. Can you give me a sense of that, or any feel for what those questions would be?

Reem Ghandour:

Sure. Yeah, so what we did is, we've added some content... And the timing is a little challenging, but it was the best we could do under the timing of the pandemic, to be honest... So, we added content around missed preventive care, and the reasons for the missed preventive care. Not just COVID happened, but: my doctor's office was closed, my kid had COVID, things like that. So, we asked questions about that. We asked content around, let me just grab it really quick, around childcare disruptions. So, we also wanted to get at that and I'm just going to check the last one...

Reem Ghandour:

Let me send a follow-up on that. I apologize. We have questions in the household pulse survey as well. So, for those of you who are familiar with the household pulse survey, so we're just pulling in those data to... Oh, I'm sorry, the last set of items are around telehealth. So, whether or not your child received telehealth, different from what you would normally receive.

Reem Ghandour:

And we tried to match the NSCH questions with what we were putting in the household pulse surveys. So, we'd have some comparison of real time of what's been happening in the last couple of months, to what actually we see once we get in the field in June.

Cynthia Powell:

Thank you.

Cynthia Powell:

And I'm told by Mia that we do have a few more minutes. If anyone else has any questions or comments before we move on.

Reem Ghandour:

I wanted to make one, if it's all right, just go back the commentary larger about how we add content, but then this, the last comment about parents access to either prenatal screening, that kind of thing.

Reem Ghandour:

One of the other things that we really look at in terms of services, when we ask questions about it, is whether or not the Bureau has a particular responsibility for ensuring access to something. So, there are any number of services we could be sort of tracking to see if parents have access to, but it's not always in the Bureau's lane to make a change in that area. And so that's one of the things we do have a fair number of items around developmental screening and things like that, but we haven't necessarily gone into the prenatal screening lane. And I think some of that is that we're still trying to figure out what is our role. And perhaps that's a unique space that this Committee in particular sits is: what is the role of the Bureau, what should we be promoting, and what are we accountable for, ultimately? Which doesn't mean we don't add things purely for the scientific, or the research sort of purposes, but we often also then track it to what would we do with those data? What are the policy and programmatic implications of the data?

Cynthia Powell:

Jeff, did you have another?

Jeff Brosco:

I guess if we have time, this Jeff Brosco. I'm following up on what you just said. Because, I also, like Natasha, was struck by the difference in reported need by parents of children with cystic fibrosis. In some ways that might be a research question, right? So, Natasha, I could write a grant and do some qualitative research, but do you do that as well? Do you sort of understand what your questions mean, follow up with some families to see. The question about unmet need is always a really tricky one, because people's perceptions of what they need might be misplaced. So, do you get a chance to do that kind of research as well?

Reem Ghandour:

That's a great question. We have not for some time. And I think to your point, yes, what you say is unmet need really depends on what you think you should be getting in the first place. And we know that wealthier parents have a different perception of what they should be getting. And so, there is a subjective nature to a lot of... I mean, that's one of the strengths of, of the survey, right? Because you're getting parents and like what they really think. And I think you always have to take a lens to say, well, why might they think that in the first place?

Reem Ghandour:

So, what I would say is that in my experience running a survey, what I've found is that we can pay for kind of standard usability kind of feasibility testing that we do with census every year. Which is like, can people actually get from point A to point B in your survey. Right?

Reem Ghandour:

And then I think, it's honestly what we found, is really almost have to put out a separate pot of money and pay someone else to do that much deeper dive.

That's sort of a true cognitive testing where you're really pulling out, what are the themes? What are you really hearing? You can do really much more targeted recruitment than you can for something...

Reem Ghandour:

And we are doing that for Healthy and Ready to Learn with the National Center for Health Statistics, cognitive interviewing lab. That was sort of by chance we had some extra money, and we were able to do that.

Reem Ghandour:

But I would argue that you probably need that kind of testing as well as the regular usability testing. And they're just different lines of investment, if I'm honest.

Kamila Mistry:

This is Kamila. Just to jump in on that. I think there's been some work on those unmet need questions, Jeff, from other surveys because those are standardized questions. And so, you may be able to... because I think they have that some deeper dives with regard to validity and really are we all thinking the same thing, those kinds of questions, I think he was referring to.

Cynthia Powell:

Joan Scott?

Joan Scott:

Yeah. This is Joan Scott from HRSA. I just wanted to note, one of the original reasons we added the newborn screening was at the suggestion of the Committee to see whether or not this was a vehicle where we could say something about the long-term outcomes for individuals and children who had been detected through newborn screening. And so, some of our initial forays into it was to see what kind of data we could get out and, is there a way that we can massage it, or improve it going

forward and how we're asking it in order to address the question that the committee's been asking for a long time about, what can we say about the long-term impacts?

Joan Scott:

Of course, the problem is that the rarity of some of the conditions that are, individually on the RUSP, it's difficult to say anything about the individual conditions and that's just our limitations. But I'm certainly welcome any suggestions from Committee members about how we can potentially look at the survey and improve our abilities to say something about the long-term outcomes for kids. Thanks.

Cynthia Powell:

Dr. Ghandour, Dr. Donney, thank you once again for sharing this information with the Committee, clearly it's an area of great interest and opportunities. So, we appreciate your being with us today.

PUBLIC COMMENT: REVIEW OF THE EVIDENCE REVIEW PROCESS

Cynthia Powell:

Next, as you know, the Committee has undertaken a review of its evidence review and decision-making processes. In February of 2019, the Committee initiated this project by convening an expert advisory panel to consider the key components of the review. In 2020 and 2021, the Committee continued to gather information on ways to strengthen the evidence review process, including: an examination of its nomination form, assessment of stakeholder values, newborn screening decision-making criteria, and the decision matrix and review of added RUSP conditions. The Committee is working to finalize the updates to these processes. And today we will have the opportunity to gather stakeholder input through public comment.

Cynthia Powell:

We received one written, and one oral request to comment on this topic. The American Society of Gene and Cell Therapies urge the Committee to collaborate with the FDA, ensure the RUSP keeps pace with treatment approvals, and ensure the process to review conditions nominated for inclusion on the RUSP is, transparent, predictable, and timely. We will now hear from Elisa Seeger who registered to submit oral comments. She has also submitted a written version of her remarks, which was disseminated to the Committee, Ms. Seeger, the floor is yours.

Elisa Seeger:

Thank you.

Elisa Seeger:

Dear Chairwoman Powell and members of the Advisory Committee for Heritable Disorders in Newborns and Children,

Elisa Seeger:

On behalf of the over 30 million Americans living with rare diseases, and as co-chair of the EveryLife Foundation's newborn screening and diagnostics working group, I am pleased to offer the following comments to inform the Advisory Committee of ongoing conversations about the review process for new RUSP nomination packages. The community congress is a forum for collaboration across stakeholders representing over 200 individual rare disease patient advocacy organizations. In addition to over 90 other healthcare and biotechnology organizations.

Elisa Seeger:

Our newborn screening and diagnostics working group is dedicated to ensuring that the rare disease community received their earliest possible access to life-saving diagnostic opportunities through newborn screening and

other diagnostic tools. Our communities work for many years with experts to develop a newborn screening system, and infrastructure, around our disease community. We invest in developing care standards and screening tools and conducting pilots, and then work to help lead the compilation of a nomination package that meets the evidentiary requirements for the RUSP.

Elisa Seeger:

We understand that the periodic review of this process necessary to ensure that standards are current and rigorous. As you proceed, we hope the review and implementation of an updated evidence review process continues to be conducted in a manner that is transparent and inclusive of patient community members as key experts.

Elisa Seeger:

We'd like to thank you for today's public comment period, dedicated to this topic. To inform your activities. We offer the following comments; as the Advisory Committee seeks to update your review process for newly nominated conditions, while also considering a new process for assessing implementation and outcomes for conditions previously added to the RUSP, it is important to ensure that any new functions of the Advisory Committee can occur without sacrificing the pace or the quality of the review of newly nominated conditions.

Elisa Seeger:

Both of these Committee roles are important to the future of newborn screening, but neither should inhibit the ability for the other to occur. As an organization dedicated to supporting patient advocacy groups that are leading newborn screening programs, we are aware that numerous strong nominations will be submitted to the Advisory Committee in the coming years. As you prepare for an increasing number of RUSP nomination

packages, we urge the Advisory Committee to develop a strategy to ensure that no condition's review is delayed due to the lack of resources to review new packages.

Elisa Seeger:

As you are aware, communities work for many years to develop newborn screening systems and develop the evidence required for a RUSP nomination. We request that the Advisory Committee considers these communities when considering any changes made to the evidence review process.

Elisa Seeger:

In addition, we ask that you consider a transition period rather than a specific transition date, for enabling a phasing of any evidentiary requirements, changes, and updates.

Elisa Seeger:

We are pleased that the Advisory Committee is requesting feedback about the educational materials that will accompany the update to the review process. These materials will help ensure the newborn screening community understands and adapts to the changes and that no community is left behind because they were unaware of the updates to the evidence review process.

Elisa Seeger:

The revisions to the review process will impact stakeholders across the newborn screening system. For these reasons, we suggest the Advisory Committee to create a variety of educational materials for newborn screening stakeholders. Identifying changes to the evidence review process, and how those changes impact specific communities throughout the entire newborn screening system.

Elisa Seeger:

Creating a collection of education materials will help to ease the transition from the current evidence review process, to the updated review process. To accomplish these educational goals, we encourage the Advisory Committee to establish a multi-stakeholder working group, to include representatives from the patient community, to inform the development and dissemination of these materials.

Elisa Seeger:

We appreciate that the COVID-19 pandemic has placed even greater demands on the precious time of our newborn screening leaders. And we are grateful for your unwavering dedication to our rare disease patient communities. The EveryLife Foundation and the membership of our community, Congress, newborn screening, and diagnostics working group stand ready to support your work. And we look forward to engaging with you over the next coming months. Thank you.

Cynthia Powell:

Thank you for your comments. Next, the Committee will break and return at 11:40 AM Eastern Time, thank you all.

BREAK

ROLL CALL

Cynthia Powell:

Welcome back everyone. We're going to start out by taking roll once again, starting with our Committee members. 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:
Carla Cuthbert?

Carla Cuthbert:
Here.

Cynthia Powell:
Kellie Kelm?

Kellie Kelm:
Here.

Cynthia Powell:
Joan Scott?

Joan Scott:
Here.

Cynthia Powell:
Shawn McCandless?

Shawn McCandless:
Here.

Cynthia Powell:
Melissa Parisi?

Melissa Parisi:
Here.

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

Annamarie Saarinen:
Here.

Cynthia Powell:
And Scott Shone?

Scott Shone:
Here.

Cynthia Powell:
And our organizational representatives, Robert Os-

Jeff Brosco:
Jeff Brosco, I'm here. I just can't find my mute button.

Cynthia Powell:
Okay. Thanks. Robert Ostrander?

Robert Ostrander:
I'm here. Power was off here all morning, but we got it back finally.

Cynthia Powell:
Oh, that's good. Debra Freedenberg?

Debra Freedenberg:
Here.

Cynthia Powell:
Maximillian Muenke? Steven Ralston? Jed Miller?

Jed Miller:
Here.

Cynthia Powell:
Susan Tanksley?

Susan Tanksley:
Here.

Cynthia Powell:
Chris Kus?

Christopher Kus:
Here.

Cynthia Powell:
Shakira Henderson? Jennifer Kwon?

Jennifer Kwon:
Here.

Cynthia Powell:
Jacob Hogue?

Jacob Hogue:
Here.

Cynthia Powell:
Natasha Bonhomme?

Natasha Bonhomme:
Here.

Cynthia Powell:
Siobhan Dolan?

Siobhan Dolan:
Here.

Cynthia Powell:
Cate Walsh Vockley?

Cate Walsh Vockley:
Here.

Cynthia Powell:
And Georgianne Arnold?

Georgianne Arnold:
Here.

Cynthia Powell:
Thank you. If I could have the next slide please. Let's see. I think I had some introductory slides that we were going to use.

NEWBORN SCREENING WORKFORCE: SHORT- AND LONG-TERM FOLLOW-UP

Cynthia Powell:
Okay. Well, while we try to bring those up. So, our last session of the meeting is going to be on the newborn screening workforce short- and long-term follow-up. The newborn screening workforce continues to adapt and evolve to challenges and new demands in the field. Thank you. It's critical for the Committee to stay apprised of current and future issues facing the newborn screening workforce. During today's panel, we will have an opportunity to explore issues pertaining to the short- and long-term follow-up workforce. We'll hear speakers spanning the continuum of follow-up services from the American Board of Medical Genetics and Genomics, the National Society of Genetic Counselors, the Child Neurology Society, and the Sickle

Cell Disease Treatment Demonstration Program. Before I turn it over to our panel of presenters, I'd like to frame the discussion with some background information.

Cynthia Powell:

Many experts and professionals comprised what we think of as the newborn screening workforce. Typically, those include genetic and metabolic specialists, primary care providers, public health professionals, which includes all public health laboratory staff and follow-up staff, additional medical specialties and hospital staff play a pivotal role as well. The current and future status of the newborn screening workforce is a key component to the success of newborn screening, from screening through diagnosis. The conversation today is a starting point for ongoing discussions, and we'll focus on genetics providers, other specialty providers, and an advanced practice provider model being utilized in sickle cell disease programs.

Cynthia Powell:

At future meetings, we hope that subsequent conversations will delve into the important role of other professionals. These include others in the public health domain, our metabolic dieticians who provide care ongoing day-to-day care for patients with inborn errors of metabolism and who are critical in devising appropriate standards of care for those patients. We also would like to include audiologists and otolaryngologists who are critical in the diagnosis and treatment for patients identified with hearing loss through newborn screening. There are many other health care professionals involved now with the care of infants identified through newborn screening, including endocrinologists and cardiologists as just a couple of additional examples. Next slide.

Cynthia Powell:

By way of recent background in 2019, the HHS Labor Appropriations Bill included language, asking the GAO to conduct an analysis of the medical genetics workforce in order to report what is known about the changes in the size of the genetic counselor and medical geneticist workforces, and what is known about the geographic distribution of these workforces. Dr. Herrera Butler of Washington requested the GAO to conduct this workforce study. The American College of Medical Genetics and Genomics, and the American Board of Medical, Genetics, and Genomics partnered to survey the field of board certified or eligible geneticists in the United States to provide information and data to the GAO. The National Society of Genetic Counselors and the American Board of Genetic Counseling provided data and information to the GAO. The GAO interviewed officials from HRSA, NIH and the Bureau of Labor Statistics. The GAO interviewed two of the seven HRSA supported regional genetics network. Next slide.

Cynthia Powell:

The GAO report shows the steady rise in the numbers of genetic counselors and a more modest trend in the number of newly certified medical geneticists, which reflects the reported average of seven genetic counselors and two medical geneticists per 500,000 individuals in the United States. Next slide. The other key findings from the GAO report are that there is wide variability in the numbers of genetic counselors and medical geneticists across States, and that most are found in hospital settings. So, what does this mean for newborn screening? Next slide.

Cynthia Powell:

GAO data represents data for both professional groups across all disciplines beyond newborn screening. We know that as we add additional conditions to the RUSP, the needs will vary greatly. I think we're faced now with the need for more neurologists. And we're

fortunate today to have Dr. Kwon talk to us more about that because that's a group that in the past has not been that involved in the short- and long-term follow-up care for children with conditions identified through newborn screening.

Cynthia Powell:

We know that as the complexity of follow-up care increases, how is that going to impact the Committee's decision-making regarding a recommendation of new conditions. If parents are going to be faced with the challenge of having to travel, perhaps thousands of miles to seek appropriate care for their child who's been diagnosed with one of the conditions. So, today's presenters will explore the challenges and opportunities that lie in the newborn screening workforce and will provide an important starting point as we explore the newborn screening workforce across future meetings.

Cynthia Powell:

With that, I would like to welcome our first presenter. Dr. Miriam Blitzer is the CEO of the American Board of Medical Genetics and Genomics. She also holds an appointment as professor in Division of Genetics and the Pediatrics Department at the University of Maryland School of Medicine and was Division Head for 20 years. She has been active in state and national work, including having served as chair of the Maryland State Advisory Committee for Inherited and Congenital Disorders. Dr. Blitzer has served on many boards, including the ABMGG and the American Society of Human Genetics. I'd now like to turn it over to Dr. Blitzer.

Miriam Blitzer:

Thank you, Dr. Powell. Can you hear me?

Cynthia Powell:

Yes, we can.

Miriam Blitzer:

Okay, great. And I really appreciate you and the entire group inviting me to participate and share a bit of data with this group. I think hopefully I'll set some foundational information for everyone to move forward. I'm going to focus today's talk mainly on clinical geneticists and I'll describe that as I go forward. Next slide please. Sorry. I was trying to change my own slides. I start every one of my tasks just to remind you what the American Board of Medical Genetics and Genomics is. Our mission is specifically to serve the public and the medical profession by establishing professional certification standards and promoting lifelong learning as an excellence as well as excellence in medical genetics, and genomics. So as a medical board, we like our other medical boards that we work with, certify individuals who have met rigorous standards to become board certified in our field and work with them to continue lifelong learning, to keep their excellence in their process.

Miriam Blitzer:

Next slide please. So, who are we? Just to give you a brief overview. We are an independent medical board. We are established 40 plus years ago. We evolved out of a need that was recognized by leaders in the field to improve the level and standards of clinical care in the area of medical genetics. And if you think back 40 years, genetics was a relatively new clinical science. It had been a research and was transitioning into the importance of a medical profession. And it was felt that starting an independent board was important in order to do two things, one to certify medical geneticists, which document their expertise in their field.

Miriam Blitzer:

This included at that time, 40 years ago, clinical geneticists who I'm going to be referring to, and these

are MDs who practice in the field of medical genetics, clinical laboratory geneticists, which at that time included biochemical geneticists, running diagnostic labs for inborn errors of metabolism and cytogeneticist, and at that time, ABMGG certified the genetic counselors as well, and with a certified individual with specialized training, defined clinical skills and expertise. The board also accredited training programs up until the establishment of the board, the residencies and fellowships were not accredited and standardized, and this was considered a really important need. Next slide, please.

Miriam Blitzer:

This evolved such that in 1991, approximately 10 years later, or 30 years ago this month, ABMGG was accepted as the 24th board into an organization called the American Board of Medical Specialties which is an overseeing organization. It's a federation of all the medical recognized medical boards, such as pediatrics, internal medicine, maternal fetal medicine, psychiatry, and neurology, and the like. So, we're the newest board at that time. We still actually are the newest board. There has not been another specialty accepted by ABMs since that time. The other thing that happened is accreditation of our programs, our residency programs transitioned over to another organization called the ACGME, which accredits all residency and fellowship programs in medicine for the other boards and ours as well. And our laboratory training programs over the last three years have transitioned over to ACGME for accreditation as well. Next slide, please.

Miriam Blitzer:

Just to give you one more background side, before I get into some of the data, I have ABMGG, that's us, that's the board. And like all medical boards, we set the standards that are required to be able to sit for the initial certification examination. We deliver and

administer the exams for these individuals. And we set the standards and administer programs for maintaining your knowledge base and continuing certification for all of our diplomates. I'm going to mention the college, the American College of Medical Genetics and Genomics, which most of you are familiar with. The ACMG is a membership based professional society. It's the professional society for medical genetics and genomics. They're the providers of educational and learning activities. They advocate for their members and fellows and the clinical profession. So, we work collaboratively. We're connected to one of other, but we are parallel on our missions and different. Next slide please.

Miriam Blitzer:

Okay. Now the board certifies individuals in clinical genetics and genomics, that's the MDs and that's where I'm going to focus the rest of my talk on. We also have a subspecialty in medical biochemical, genetics, where individuals who are clinical geneticists can get additional training and focus their practice, which is a very important for this group on inborn errors of metabolism and certainly all those disorders that are identified through newborn screening. We also certify laboratory geneticist in the diagnostics labs for biochemical genetics and the specialty of laboratory genetics and genomics, which is a merger between cytogenetics and molecular genetics. Next slide.

Miriam Blitzer:

The residencies, and I bring up how we train our MDs very briefly because I think this is where the workforce challenge comes in. To become a medical geneticist or a clinical geneticist, you have to complete a two-year accredited residency, which requires additional pre year of residency in another field with a 12 months of direct patient care or you can complete what we call a combined residency.

Combined residencies are approved by two specialty boards, like American Board of pediatrics, American Board of Medical Genetics, where we have approved combined training for four years. And at the end of that training, the individual, the physician is able to sit and become certified in two specialties. And these are the different areas we have. This has been actually a way we are seeing an increase in a workforce. Next slide.

Miriam Blitzer:

I'm just going to say that we have this many accredited programs, 46 around the US, that's a relatively small number. And the biggest goal that we have is to recruit more clinical geneticists into these areas. Next slide. I want to sort of ask you to look at the far right. These are residents. We're looking at the pipeline here, which are very important components. And if you look at the far right, it says how many residents are in training programs. From our standpoint, there's been a significant increase from '14, '15 to this current year from 129 to 161. We're not talking large numbers, that's a large increase. But when we think about the numbers, they are relatively small. Next slide.

Miriam Blitzer:

This is a slide with little bit more detailed than the one that Dr. Powell showed from the GAO report. We provided directly from the board a fair amount of data to GAO. And this is showing the numbers of newly certified clinical geneticists in the last 10 years. And are we seeing an increase? Yes, we are. We're hoping that this will continue. We give our exams every other year, so they are given in two months from now or three months. Our numbers are still small because these are every other year exams, but we are starting to see an increase. Question is, is this insignificant enough at this point? And then how long will this take? Next slide.

Miriam Blitzer:

I know these slides have been provided to you, but basically this is at the top line. It says that there are currently sort of 1,240 clinical geneticists in the US. These are practicing, taking out retired, taking out deceased, and these data will be updated. This is the data we submitted to the GAO. This will be updated this year. This will likely increase. If you look at the bottom from our inception as a board, what you're seeing here is the clinical geneticists in practice in red compared to those that were certified that year. And you'll see that as expected from the early years, a fair number of individuals have retired or have died. And we are starting to see a maintenance of clinicians going forward. And obviously this is going to depend on maintaining and seeing how we're going to increase these numbers. Next slide. I'm going to skip this slide which just show some of our demographic breakdown and show it a little differently on the next slide, please.

Miriam Blitzer:

This shows just where we are right now with non-retired clinical MD geneticists who we know are in practice. And just to give a hand to, because I think looking at our workforce and the big focus for the ABMGG in the next few years is going to be defining our workforce in much more detail. And working with our professional societies on recruitment. This is not really surprising for those of us in our field. We actually are a predominantly female majority; I should say female for clinical geneticists. We are not very diverse. And this is a concern of ABMGG and our specialty as it is for any medical specialty at this time. And this is an area that we are beginning to focus on. And we are also right now looking at the data for our pipeline, our trainees in comparison to our diplomates now. And it also impacts, and we're also looking at scope of where people are practicing. Next slide.

Miriam Blitzer:

So, Dr. Powell mentioned this, and this is just an updated graph of what we had submitted to the GAO as far as where our clinical geneticists and how certainly are they going to be able to provide services, especially for follow-up of newborn screening, depending on where they're located. Not a surprise to us in the field, but also a concern is if you look, there is a number of states that have one, two or only three geneticists for their entire state. And obviously there is a concentration in particular areas and certainly in the major academic centers, as Cynthia mentioned. And we have one state, as you can see with no clinical geneticist practicing currently in that state, at least according to our last run of these data. We'll rerun these data again after our next certification exam and it will be a bit more specific. Next slide.

Miriam Blitzer:

So as Cynthia also mentioned, the ABMGG and the ACMG did a workforce study in response to the GAO. The data the board provided the GAO was not through the survey, it was complete data. Survey data always is a little more difficult or challenging to interpret, but these data were published a week and a half ago on May 3rd in Genetics and Medicine. In this survey, there were almost 500 clinical geneticists who responded. And the data I think were fairly telling in the sense that the average workweek was 50 hours, and we went through in this looking at the wait time for new patients, which is significantly long, location and I showed you the map and the amount of clinical duties.

Miriam Blitzer:

I will say that telehealth and telemedicine has been helpful in genetics. And we were able to sort of manage the COVID pandemic a bit better in our field, but certainly the duties are really challenging here,

especially with newborn screening in the sense of looking at how do we get patients in because there are emergency patients. Conclusions from this are very straightforward that there's a gap between genetic services needed at the workforce capacity and this is where the concerted effort really needs to be made. Next slide. I think that is my last. Dr. Powell. I don't know if we're holding questions to the end, which I am guessing.

Cynthia Powell:

Yes.

Miriam Blitzer:

I welcome to answer.

Cynthia Powell:

Thanks very much Dr. Blitzer. And we were going to hold questions and have a general discussion after all of our speakers have presented if that's okay with you.

Miriam Blitzer:

Okay.

Cynthia Powell:

So next we will hear from Cate Walsh Vockley who serves as the Committee's organizational representative from the National Society of Genetic Counselors. Ms. Walsh Vockley is a senior genetic counselor at UPMC Children's Hospital of Pittsburgh Division of Medical Genetics and is certified by the American Board of Medical Genetics. Her current clinical interests include genetic counseling for inborn errors of metabolism, especially newborn screening disorders and working with the plain communities of Western Pennsylvania. Other interests include genetics education and the ethical implications of clinical genetics and genetic testing. And I'll turn it over now to Cate.

Cate Walsh Vockley:

Great. Thank you, Dr. Powell, and thank you to you and to the Committee for bringing this topic to conversation. I think it will be so essential as we continue to add to the rest. My conversation is perhaps a little bit more applied and a little bit more specific to newborn screening as we look at genetic counselor workforce and needs. Next slide please.

Cate Walsh Vockley:

So next slide. I thought it was important to start our conversation by looking at what roles genetic counselors do currently play in the field of newborn screening. We actually do a professional status survey on an every two year basis through the NSGC. And while it lists a very small number of genetic counselors in newborn screening and in public health, I think that reflects the fact that many genetic counselors have newborn screening as a part of their work during their activities that not too many genetic counselors are only working solely in newborn screening because I've had many conversations over the last two months with people who don't see themselves as primary newborn screeners.

Cate Walsh Vockley:

So, lots of under ascertainment of the workforce, I think there. But if you look at genetic counselors who fill roles in the newborn screening system, we have genetic counselors working everywhere from the federal genetic services programs through the regional genetics networks that they've been mentioned several times during this meeting, coordinators of state genetic service programs, serving on state newborn screening advisory boards, patient advocacy programs, a lot of genetic counselors working as laboratory liaisons through the private and state run, newborn screening laboratories, many genetic counselors who work with

families who are referred because of a positive newborn screen, both for short-term and long-term continuity of care relationships. We also have genetic counselors who work in clinical research and clinical trials for disorders that are identified through the newborn screening programs, and also some who work supporting therapeutics for patients who are identified. So really across the spectrum of the newborn screening system. Next slide please.

Cate Walsh Vockley:

So, I'm going to focus on some workforce challenges for genetic counselors and newborn screening. Next slide. A lot of these will focus on educational needs for the various people involved in newborn screening. Certainly, families need to be informed about new diagnoses or new potential diagnoses, other healthcare professionals who are not familiar with these disorders. And even the newborn screening workforce in the laboratories may need to be informed about newly added conditions. We certainly need knowledgeable informants to work with families, what we call families in waiting. People who are identified depending upon the state screening protocols may have a positive biochemical screen and need to have follow-up testing, certainly confirmatory testing at a minimum. Families may have a significant wait times and may have many questions. The addition of Pompe and MPS 1 have already significantly increase the need for individuals knowledgeable about these disorders and about the molecular genetics, not only of the genes involved, but pseudo-deficiency alleles and similar issues.

Cate Walsh Vockley:

Cascade testing has also become a big issue for conditions as they're being added, particularly in the early timeframe for conditions like Pompe, ALD and SMA, because there may be family members who are young

enough yet to perhaps be affected by one of the later onset variants who are candidates for screening. Diversity, as Dr. Blitzer mentioned for among our providers is an important issue as well as we serve a more diverse and complex population seeking care. And this impacts not only clinical care, but also public health with a variety of different diversity issues. And then as I mentioned, we need individuals who are knowledgeable about clinical trials for conditions on the risk as more and more therapeutics are being developed for these conditions.

Cate Walsh Vockley:

More and more therapeutics are being developed for these conditions. Next slide please. Another issue that came up frequently as I talked to my colleagues was one of the needs for competitive compensation based on training, experience and skills. And this impacts both public health and clinical care. There's a lack of defined positions for genetic counselors in public health. Oftentimes genetic counselors are taking coordinator positions or administrative positions that don't acknowledge their degree and their skill level. This affects not only retention of more experienced counselors, but even entry level positions where it's difficult to recruit knowledgeable individuals because of salaries that are not competitive. There's also a need for promotion opportunities or a job ladder. There are also lack of defined civil service positions for genetic counselors. We have acknowledged in all of this conversation that state programs are certainly under-resourced, but we need to look at more creative ways to fund these positions so that we can compensate people for their skillsets. And all of this has impacted, of course, by the limited a number of MD geneticists, certainly those in biochemical genetics to do the follow-up work that these families need. Next slide please.

Cate Walsh Vockley:

Just a little bit more information on some of the salary issues. If you look at again, small numbers with this survey, we had about a 50% response rate and only a small number of newborn screening and public health. But if you look at median salaries for counselors who identify as working in newborn screening and public health, as compared to all counselors reporting, there's a significant difference in compensation that we would like to see improved. Next slide please.

Cate Walsh Vockley:

So looking at present and future solutions, genetic counselor activities and newborn screening. One thing that some programs have been doing is looking at increasing prenatal newborn screening education for families. We had a pre-conference symposia around the NSGC meetings a number of years ago targeting prenatal genetic counselors who discus screening throughout pregnancy and we hope can integrate inclusion of newborn screening education in the prenatal genetic counseling setting. Time is an issue for these people because they have lots of different things to talk about.

Cate Walsh Vockley:

They've also been a number of innovative solutions for newborn screening education that are being developed. Some of our students particularly have been very creative with chatbots and iPhone applications, and there are many videos developed or in development that have been done by genetic counselors. Hawaii, if you're not familiar with the materials available there, this is a project of the Western States Regional Genetics Network, not NSGC specifically but involved in quite a number of genetic counselors. They have developed parent fact sheets and are in the process of developing new videos all done by genetic counselors in conjunction with specialty care providers and the link there will take you to those. If you look at the ALD

one in particular, that one has a new video that's just been developed. Next slide please.

Cate Walsh Vockley:

We also have information from some of our colleagues about families expressing frustration about notifying providers not always being knowledgeable about the condition in question. Certainly, genetic counselors have training and expertise in delivery of high anxiety news that can be very effective in that setting. As more and more molecular results are being included in some states with the first notification of results, families are asking for explanations about this, as are our primary care providers who may choose to inform the family but have a need for some background information that they may not have had in their training before communicating with the families and in many areas, genetic counselors are providing some of that information to primary care providers. There are also genetic counselors who play a role in helping to do risk assessment and look at needs for immediacy of referral. As conditions with broad phenotypes are added to the RUSP and the state newborn screening panels. Next slide please.

Cate Walsh Vockley:

Genetic counselors are also being asked to provide ongoing support for families. Not all clinics have... Most have nurses involved, not all have social workers as part of their teams and genetic counselors often fill multiple roles, including genetic counseling, care coordination and the cascade screening that I mentioned earlier, looking at family members who may be at risk. As the lysosomal storage disorders are added I think this has become a real issue and may continue. Other genetic counselors that I've talked to play a role in quality improvement and quality assurance within their state systems working to decrease false positives as we learn more about screening for some of the lysosomal

storage disorders, and also play a role in providing follow-up data on short- and long-term follow-up for state systems so that assessments can be done to look at allocation of resources for family needs. Next slide please.

Cate Walsh Vockley:

Much like what Dr. Blitzer talked about, there has been an increase in the number of genetic counselors who are being trained. There's been an exponential growth in training programs in the last five years. And we now have 55 total programs with a student enrollment increase by 52% and a professional growth rate of over a 100% in the last 10 years, and it is anticipated that this continues. There are now over 5,000 practicing genetic counselors, while not all of them are in newborn screening obviously, that number is growing as well. There has been an increase in metabolic and newborn screening curriculum content in some programs. And also, there are development of CEU materials for previously trained genetic counselors in newborn screening and metabolism. There also is a long-standing focus on public health in the University of Pittsburgh program. So, students are being trained in public health as part of their genetic counseling training. Next slide, please.

Cate Walsh Vockley:

Our focus in diversity, we have a strong commitment on improving the diversity in our field. The organization commissioned an assessment by an external group, The Exeter Group, looking at current diversity issues and are in the process of developing a five-year organizational plan. As has already been mentioned, NSGC also is contributing to the genomics workforce diversity initiative. And there are several programs in place already that are very effective. There's, again, Western States Regional Genetics Network has been active in a minority genetic professional network. And

there are 800 current members that support and mentor minority students into the field and support those already in the field. There's also a newer program out of Arcadia, now University of Pennsylvania training program. Erica Price, who is a recent graduate, developed the golden program, and that also supports young minority students who are interested in the field of genetic counseling. Next slide, please.

Cate Walsh Vockley:

As far as the compensation goes, and I'll just go through this briefly, NSGC has had a number of legislative efforts to increase access to genetic counseling services. House Bill 2144 currently before the House of Representatives and Senate Bill 1450, just introduced about a week and a half ago, look at GC reimbursement for services for Medicare and CMS. And while this may not seem relevant to newborn screening because it doesn't address pediatric reimbursement necessarily, we believe that there will be downstream effects because hospitals will be able to bill for genetic counseling services, allowing them to hire more genetic counselors, allowing genetic counselors and private practice to bill. And also this may encourage other covering entities like Medicaid and private payers to pay for genetic counseling services, thus increasing access to genetic counseling services. Next slide. I have a couple of supplemental slides. Next slide.

Cate Walsh Vockley:

First, I'd like to thank some of my colleagues who contributed to the presentation. Next slide. This is just ABGC diplomats by year from 1982, through current. As I said, over 5,000 -- 5,629, currently board-certified genetic counselors. Next slide. A representation of the output from the Exeter Group is a JEDI assessment that you can review at your leisure. And then one more slide I believe is the link to the

information from the Western States Regional Genetics Group patient, or family education brochures, and they are really comprehensive and very nice. And as I said, people are welcome to utilize them and the YouTube videos to share with their patients as long as acknowledgement is given to the team that produced them. And I'll stop there and wait for questions at the end. Thank you.

Cynthia Powell:

Thank you, Ms. Walsh Vockley. Next, we will hear from Dr. Jennifer Kwon, who serves as the Committee's organizational representative from the Child Neurology Society. Dr. Kwon is an academic child neurologist with a strong interest in improving long-term clinical outcomes in children diagnosed with rare disorders by newborn screening. She is currently at the University of Wisconsin where she is the director of the pediatric neuromuscular program at the American Family Children's Hospital, which is a muscular dystrophy association care center, and a parent project muscular dystrophy certified care center. Dr. Kwon.

Jennifer Kwon:

Thank you. I'm Jennifer Kwon, and I'm happy to speak as the organizational representative for the Child Neurology Society to the Advisory Committee. Next slide, I am at the University of Wisconsin and my only disclosure is that I'm the site PI for Novartis, formerly AveXis, sponsored clinical trials in gene therapy for spinal muscular atrophy.

Jennifer Kwon:

Next slide. I'll be talking about the Child Neurology Society, briefly introducing our society as well as who we are as child neurologist and how we've grown. Despite our growth, we still have a number of workforce challenges primarily to keep up with the new discoveries in the field and the new treatments. We do

need more child neurologists, as well as child neurology sub-specialists, including neuromuscular sub-specialists and those who specialize in other degenerative conditions, such as inherited white matter disorders. The Child Neurology Society in the past year and a half has come up with some innovative solution to addressing the challenges that we face as a profession. So I'll briefly talk about our COVID response and tele-health guidance, as well as ways we have of trying to increase recruitment over time.

Jennifer Kwon:

Next slide please. So, the national office of the Child Neurology Society is based in St. Paul, Minnesota, and our executive director is Roger Larson. Our current president is Dr. Phil Pearl. The Child Neurology Society was founded in 1972, and it had an initial enrollment of about 200 members. Today the membership is over 2000 and it exists to serve academic research and practice-based needs of child neurologist. So, who are child neurologist?

Jennifer Kwon:

Next slide please. So, after medical school, child neurologists undergo a five-year post-graduate residency. Historically, child neurologists were actually adult neurologist who had a particular interest in neurologic conditions presenting in children. Our training reflects that background. So, we have two years of training in pediatrics, a year of adult neurology training, as well as two years dedicated to pediatric neurology. Many of us go on to additional subspecialty training after that, either in epilepsy, neurophysiology or neuromuscular disorders. Because of our year training in adult neurology, many of us also see adults actually in practice or continue to follow our pediatric patients into their adult years. Today, there are over 70 university-based child neurology training programs, enrolling over 150

candidates yearly in the US and Canada. That's an increase from 2000, so over the past 20 years, when there were only 58 programs and approximately 80 positions. So, that is one. We've had workforce concerns since the late 90s and this was one way of responding to those concerns. Also, it's helped that we are part of the Match Program.

Jennifer Kwon:

Next slide, please. I really enjoy Dr. Blitzer's numbers from the board. I don't have those similar numbers available to me, but just based on child neurology society membership, obviously our numbers have grown. So basically, as I said, we now have over 2000 members and you can see how that's increased over time. Unfortunately, it's still not enough to meet the demand that we have right now, and that's not even including newborn screening. So next slide, please.

Jennifer Kwon:

The reason for that demand is really just the dramatic increase we've seen in medical knowledge, including information about genetic mechanisms and how that leads to novel treatments. More and more of us are seeing the need for sub-specialty training because standards of care for some of these rare conditions, so I'll give you an example, infantile spasms, or even some of the disorders that we see in newborn screening such as ALG, they have standards of care that we feel in order to really implement require a higher level of training than we received in our general child neurology training. So having that additional specialized experience can be helpful.

Jennifer Kwon:

Even community-based general child neurologists find themselves increasingly facing rare conditions for which they need to keep up with this new knowledge. And, again, having access to specialized centers is

helpful for our community-based colleagues. So, because of the complexity of our patients, we find that we routinely engage with other specialists and services. We care for our patients in a multidisciplinary world filled with therapists, geneticists, genetic counselors, and more. And that also impacts what we're able to do and how we're able to provide services. So, all of these things put a lot of pressure on our field. Next slide please.

Jennifer Kwon:

So, as I said, we need more child neurologists to keep up with these innovations and the heightened expectations that we have. We also have an aging workforce that we need to replace. Even though the number of training opportunities has increased, really we haven't seen much of an increase in terms of interest. So even though we have twice as many people applying for available residency slots, the overall number of residency applicants has not really changed over the years. So, we do need to think of ways to improve recruitment of new trainees. Part of the issue that we face is that compensation has not been good as it has been for adult neurologist. Child neurologists, we span that area between loving the brain and neurology and also always wanting to experience the joy of working with children. And so, part of the cost of that is that our discipline takes a lot of time and we're not paid at the same rate as our adult neurology counterparts.

Jennifer Kwon:

We do tend to work in large urban areas. And I actually think that pressure is going to increase as our patients become more specialized and we know more about genetic mechanisms and ways of intervening with care, child neurologist feels more compelled to be at centers that have the kind of imaging services and neurodiagnostic services that they need to be able to

provide good care. So, we tend to cluster in more urban areas, and that is certainly a stress on our patients because they need to travel longer distances to see us. We've tried to work out different ways of addressing that. Next slide, please.

Jennifer Kwon:

So, on the top right is the fact that since COVID we've been using more multidisciplinary tele-health services, but I would say that over the past 10 years, child neurologists have really seen the need for tele-health expansion. There just have been a number of barriers. The one positive about COVID that was brought up at yesterday's session, I think Michele Caggana, was the first one who mentioned it, but other people also chimed in to say that the increasing use of tele-health has been a real benefit of the COVID pandemic. And I would say that we've experienced it as well. Many of the state-based barriers and insurance space barriers to tele-health disappeared under the emergency acts of COVID. And we were able to see patients much more readily without having to force them to travel to see us. And so that's been a real benefit. And unfortunately, we're seeing that disappear as states no longer are bound by emergency procedures and are not really allowing telehealth services, for example, across state lines, that's been a real problem.

Jennifer Kwon:

What we did also do during COVID is that when patients would come to see us for their in-person visits, we tried harder to increase our care coordination for those patients. So, if I knew that a patient had to come in for an NJ tube change, for example, we would try to add on some surveillance, laboratory testing and other visits during those in-person trips by families. Again, that increase in care coordination is a burden that child neurologists take on. We especially took that on in the neuromuscular program for our patients

who had been identified by newborn screening, let's say with SMA, who needed to come in for a visits for lab work. The other response that child neurologists across the country are implementing, those of us who are in larger centers in more urban areas do try to travel ourselves to outreach clinics to see patients in more distant parts of our states.

Jennifer Kwon:

So, during the COVID times especially, the Child Neurology Society came up with a number of statements and practice guidelines to help child neurology members. So, that included an ethics statement about accessibility to services during the COVID-19 era. We had a change in practice parameters for infantile spasms, again, to allow families and providers alternative ways of treatment at a time when it was harder to see patients in person. There were guidance's about how to conduct tele-neurology examinations and I will say that one of the positives of telehealth visits is that it's often a much more comfortable visit for children, as you can imagine, they're playing in their home and you can see much more active engagement sometimes doing home visits that way.

Jennifer Kwon:

There was also guidance from the Child Neurology Society about reopening practices and how to safely do that, as well as a statement on institutional racism, which is an obvious public health concern. Child neurologists, because of the conditions that we treat, do depend on our society to help us understand the best care practices, how to safely care for children who have rare conditions. And so, we we've benefited from the SMA newborn screening guidelines that were sponsored by Cure SMA, but included the input of many child neurologists. The X-linked adrenoleukodystrophy guidelines likewise, have been a great help in terms of following children with X-ALD and I look forward to

looking at the guidance and the patient information that Kate Barkley had noted in her talk. Next slide please.

Jennifer Kwon:

So, SMA was added to the RUSP in 2018, and this map shows the number of states that have adopted screening for SMA, 36 as of last month when I looked at the map and that's about 70% of newborns in the US who are being screened for SMA. And the reason I bring that up, next slide please, is because, of course, SMA newborn screening has I think really brought child neurologists into the fold of newborn screening, short and long-term follow-up care. So, we are the primary specialist who see infants identified by state newborn screening programs. Before there was newborn screening for SMA, we were the ones who led efforts to define natural history studies that could be used in the clinical trials that help develop the novel treatments that are available for spinal muscular atrophy. I think that it's hard to express how overwhelming it has been to see the benefits of newborn screening in this population.

Jennifer Kwon:

So, I would say that one of the new things that we'll have to do, so short-term follow-up for SMA has involved a fair amount of just scrambling to identify the appropriate treatment and to institute them early. And you may wonder what long-term follow-up needs there are, but I think that in the 2018 meeting, before it was approved on the RUSP, there were a number of concerns that were raised about these highly efficacious treatments, but the fact that we hadn't really been following these treatments for very long, we were only seeing the data over a few years. And how do we know about how long and how durable these treatments are? So, I think most child neurologists are committed to following these patients who are referred

soon after newborn screening. We're committed to following them for years in terms of understanding the natural history of SMA that's being treated, or that has been treated in the pre-symptomatic stage.

Jennifer Kwon:

So, we have always recognized the importance of multicenter collaborations and I should also say the importance of multidisciplinary collaborations. While SMA is a particular disorder that I think most child neurologists are fairly comfortable with, the other disorders that we're going to be involved with that are identified by newborn screening are ones where we would certainly need multi-disciplinary help. I think Cate Vockley brought up the comment about patients in waiting in her talk. And I was just thinking about how that really applies to children who identified with late-onset Pompe disease and with adrenoleukodystrophy. I think they feel like they're waiting for the shoe to drop. In terms of being child neurologist and wanting to follow their neurologic status, how do we bring in the other disciplines who were also needed to care for these patients and to answer these families' questions?

Jennifer Kwon:

So, while we've been energized by the benefits of SMA newborn screening, we do have a lot of questions that we have about what our role is and how to make the most of our role with these other newborn screening programs.

Jennifer Kwon:

So, this was really a fairly brief talk, next slide, to introduce you to who we are as child neurologists and what we want to do to help with future newborn screening program efforts. I want to thank Margie Ream, who is a child neurologist at Nationwide Children's Hospital. She's been involved with the Evidence Review Group. And I think that the involvement of child

neurologists in the Evidence Review Group, as well as on this Committee, will be very important as new disorders are being introduced to the Committee. I also wanted to thank my colleague, Adam Wallace, a child neurologist here at the University of Wisconsin, who has also thought a lot about workforce issues and how to improve that. And really the key is to improving medical school enrollment overall in our high school and undergraduate, and then improving the exposure of neurology to our medical students. My last slide was just a slide about questions, but I'll wait until the end. Thank you very much for your time.

Cynthia Powell:

Thank you, Dr. Kwon. Next, we will hear from Dr. Julie Kanter and Dr. Marsha Treadwell, who are both principal investigators for the HRSA funded Sickle Cell Disease Treatment Demonstration Program. Dr. Julie Kanter currently at the University of Alabama at Birmingham is a lifespan hematologist specializing in sickle cell disease. She works closely with a variety of national partners on advocacy and research, as well as industry and federal partners to develop new treatments for sickle cell disease. She has specific interest in gene therapy, adhesion therapy and novel combination therapy. Dr. Kanter has received NHLBI funding in dissemination and implementation to improve access to care for affected individuals.

Cynthia Powell:

She has authored and co-authored more than 100 articles, book chapters and abstracts. Dr. Treadwell is a professor of pediatrics in the division of hematology at the University of California, San Francisco. She is the Jordan fund endowed chair in the Department Of Hematology, Oncology at UCSF Benioff Children's Hospital, Oakland. Dr. Treadwell is co-PI and regional director for the HRSA-funded Pacific Sickle Cell Regional Collaborative that focuses on improving access

to sickle cell disease care in 13 western states. She is co-PI and director for the Sickle Cell Care Coordination Initiative funded as a Sickle Cell Disease Implementation Consortium Project by the National Heart, Lung, and Blood Institute. Dr. Treadwell is recognized internationally as an expert in the transition from pediatric to adult care, quality of life, patient reported outcomes and community engagement. Dr. Kanter, I will now turn it over to you.

Julie Kanter:

Thank you very much. And thank you for having both of us today. I would be lying if I said that these efforts were just mine and Dr. Treadwell's alone. In fact, we represent two regions of the HRSA Sickle Cell Disease Treatment Demonstration Project, and all of these efforts have really been done in collaboration with our national groups, all our national PIs. So we're going to talk about increasing access to expertise in sickle cell disease care by training of...

Julie Kanter:

Talk about increasing access to expertise in sickle cell disease care by training of advanced practice providers. Next slide. These are disclosures which really aren't relevant. Next slide. So, the goal, it seems in many genetic diseases is too many patients, not enough experts to really help manage the care of these individuals. And this is especially true in adults living with sickle cell disease care. And in fact, many adults are receiving care in settings that don't have appropriate resources. I can tell you stories that are amazing. 40-year-old patients who've never seen a sickle cell specialist, of 23-year-old patients who once saw sickle cell specialists when they were one or what they remember as one, and then haven't seen one since. And so, the goal all along has been, how do we improve access to care? And in previous

sickle cell disease treatment demonstration projects, this was thought to be through primary care providers.

Julie Kanter:

If we could further educate primary care doctors in managing sickle cell disease, that would increase the number of physicians who had expertise to provide this care. However, despite multiple years of effort, we've really recognized that there is a concerted lack of interest. Now, primary care doctors are heroes in our field of medicine. They're managing every condition out there and they don't often have the time to dedicate to learning enough about sickle cell disease, a multi-system disorder to provide this kind of expert care. So, there's lack of interest due to lack of time. And there's clearly insufficient reimbursement. There's also insufficient support systems for primary care doctors, social workers, case managers, and other necessary personnel. So, we attempted a new solution during this last sickle cell disease treatment demonstration project grant. It was called the STAMP initiative, the sickle cell disease training and mentoring program for primary care providers.

Julie Kanter:

And it was enjoined project of the grantees and the office of minority health and OMH recruited primary care providers from everywhere, multiple societies centers, et cetera. They really said to us, "We don't believe you guys have tried hard enough to recruit these primary care doctors." "We're going to try for you". And well, over a thousand individuals were recruited. In my state alone it was over 300 individuals and the R part of this, the regional grantees were to hold ECHO education and mentoring sessions designed for primary cares doctors.

Julie Kanter:

The good news is, STAMP was well attended, but it was really a variety of healthcare workers and community members, was very few primary care doctors, very few primary care doctors who did attend didn't reach out and attend more than one session. And actually, none of them followed up with an ongoing interest to manage or take on the care of patients with sickle cell disease. So obviously we said, "Okay, we have to find another way." So, we regionally, all of the PIs had gotten together and we collectively believe it's appropriate to shift capacity to building advanced practice providers who can be trained to work as extensions of sickle cell specialists. Next slide.

Julie Kanter:

So, from there was born the SUPPORT program. The sickle cell disease, advanced practice providers, opportunities, resources, and training program, known as SUPPORT because it's made way too hard to say all of that at once. And this is a collective program that really is run by all of us. So, Marsha and I are representatives of a greater good. We're really doing a lot of this work. And that's especially the synergy region Northeast, run by Dr. Sophie Lanzkron.

Julie Kanter:

And there are three main components of this program. So first there's an APP focused ECHO. This is an ECHO that's recurring telementoring session, and it's specifically geared for APPs. In fact, it's run by and sponsored by advanced practice providers with physicians, just lurking in the background in case they're needed. It's case-based learning and mentoring sessions with brief didactics on specific areas of sickle cell care. And again, fully facilitated by APPs.

Julie Kanter:

As many of us already have specialized APPs in our clinic, they are really the best ones to teach other

peers. And they're open to all APPs regardless of their expertise in sickle cell disease care or the participation in this second component. The second component is a web-based online curriculum. This is a special learning management platform that we have developed to specifically allow for asynchronous education of APPs in sickle cell disease care.

Julie Kanter:

And this is a series of, as you might imagine, curriculum where they have pre- and post-tests, and if they complete everything in addition to their ECHO attendance and the following observership, they actually will get a certificate. So, the observership is at sickle cell disease centers of excellence. And these are times that APPs can come and observe and really get additional training in quality evidence-based care for sickle cell disease. So, what we've done is all of our treatment demonstration project grantees have identified and referred APPs to participate in SUPPORT. We've all worked together to formulate and provide the curriculum content, provide the observership opportunities, and then to collectively pay for this as well out of our treatment demonstration grants and OMH supplements. Next slide.

Julie Kanter:

So, what are the goals? To increase access to quality evidenced informed sickle cell disease care. We know that we need more providers, and we are partnering with APPs to do this. So, partner with site leads and in specific States to train APPs, and then embed those APS in primary or secondary settings. So that may be for example, that we have more APPs within our sickle cell disease centers, so that we can increase the number of patients that we manage, but it may be, as I did in South Carolina, that I have an APP in an entirely separate area of South Carolina, who really runs the day-to-day clinic of a small clinic with sickle cell

disease patients. And that I am there constantly if she needs me or to help manage those patients and to provide opioid prescriptions, or it could be that an APP works in primary care but agrees to take on a significant number of individuals living with sickle cell disease.

Julie Kanter:

We're including these ECHO sessions that are tailored for APPs, we're increasing the accessibility to specialists for on-demand consultation and then facilitating additional co-management so that we can really get hands-on and improve the care for our patients. We're doing measurement in many different ways. We're measuring the number and type of participants in the ECHOs and other trainings. The number of individuals with sickle cell disease served and then looking at other things such as hydroxyurea prescriptions, pediatrics who have completed transcranial doppler or stroke screening, and are they getting the right immunizations? Next slide.

Julie Kanter:

So, this is our logo, our SUPPORT logo. Again, it's a one-year program and the participant is required to attend the ECHOs, complete the curriculum and an observership. There's many modules that make up this curriculum, that includes benign hematology, pain management, which you can imagine we do quite a lot of, especially in adults with sickle cell disease, but also with children. Identifying, recognizing, and treating acute sickle cell disease complication. Understanding the use of disease modifying and curative therapies and ensuring that we are appropriately evaluating and helping those with specific issues with social determinants of health.

Julie Kanter:

As discussed previously, there's also modules on implicit bias and addressing systemic racism. Next slide. So, this is just our ECHO flyer that shows you what we're doing and how our goals are really to make hub and spokes using our SUPPORT ECHO series. And we've had excellent attendance. So, we're very excited about that. Next slide. And with this, I'm going to turn it over to Dr. Treadwell. Thank you very much.

Dr Marsha Treadwell:

Thank you so much. And thank you to the organizers for having both of us. As Julie mentioned, we definitely are a part of a big collaborative that's national. We represent two of the regions, but there are five regions for the sickle cell disease treatment demonstration program. So, I'm just going to talk a little bit about our evaluation, which has really just begun. We're really working with a supplement right now. And so, with that, the regional collaborative, the five regions are as Julie mentioned, we are looking at these advanced practice providers and how we're going to evaluate that is, their assessment of their sickle cell disease knowledge based on existing guidelines and their confidence in their knowledge of sickle cell disease.

Dr Marsha Treadwell:

Within the Pacific Sickle Cell Regional Collaborative, we're also using some implementation science strategies to evaluate this program. So, I can talk a little bit more about that. We have... So, we have two APPs in the Pacific region. We have two hematologists who are mentoring them, and the mentors completed a practice change readiness assessment measure. And then our study coordinator here interviewed both mentors and that the advanced practice providers, the two advanced practice providers to find out what they feel is needed to best support their training, what will be needed to extend this training beyond the supplemental pilot and

anticipated challenges, as well as other thoughts. Next slide.

Dr Marsha Treadwell:

So, the two sites in the Pacific region, one is pediatric, and one is adult. With the pediatric site, the hematologists and the advanced practice provider are at different hospitals. The mentor's practice size is about 40 patients with sickle cell disease per month. They're just getting started together over the last few months. So, they haven't really co-managed patients yet. Where we're starting in terms of our metrics is that hydroxyurea adherence is quite high as is the adherence with the transcranial doppler screening. Site two is an adult site. So, the APP is actually in a primary care clinic that serves sickle cell disease. The mentor's practice sizes about 15 a month. So, it's smaller than the pediatric site. The mentor and APP are currently co-managing. So they came into the project co-managing people with sickle cell disease. With the hydroxyurea adherence and immunizations, not quite as high. Next slide.

Dr Marsha Treadwell:

So, when we asked in terms of baseline readiness, the pediatric site really focused on in thinking about how ready they are to implement the advanced practice training that this training really fit in with their organizations, strategies towards innovation, it's aligned with the institution's priorities. And they really feel that their ability to observe results is going to really help move the project forward. They also felt very strongly that there is a project program champion within their organization, good relationships, the ability to use evidence informed care and the areas to improve might be, there's not really a systematic place to... System in place to monitor how well the practice changes end up being implemented. They're concerned about a return to the previous way of

administering care and that this the priority in terms of sickle cell may not be as high as it should be.

Dr Marsha Treadwell:

Next slide. For site two similar ratings in terms of that, this is an innovative program and so that's attractive to the larger institution. And again, it aligns with the larger institution's goals. They do have a champion... Again, very similar to the pediatric site in the remaining strong ratings of readiness, as well as the weaker ratings. Next slide.

Dr Marsha Treadwell:

So, in doing our qualitative interviews, areas of consensus between the mentors and the mentees across the two sites, they all felt that the APP project was a very important strategy for increasing access to quality evidence informed care for people with sickle cell disease, with a representative quote that prior to this, so many things were falling through the cracks. They did have some concerns about communication within and between health systems across the two sites and across the mentors and mentees, that there's a need for a broader staff education about sickle cell disease, not just having it lie on the APP and the mentor. It's important for the mentor and the APP to have a very collegial relationship with easy and open communication, that this funding supported the supplement is allowing this project to unfold. And what would program success look like to be able to intelligently and confidently speak about sickle cell disease versus having a superficial knowledge? Next slide.

Dr Marsha Treadwell:

Site differences in one of the sites, the APP again is dedicated to the sickle cell population. And the other they have their duties. In the pediatric site the relationship between the mentor and APP is new, whereas

in the adult side it was already established. It was mentioned in another presentation here that access to a social worker is so important for a genetic condition, sickle cell disease no less, and that access is limited. And that... One of the sites it was limited. And then one of the sites as well, there was some concern expressed that other providers might feel that the APP was actually taking their patients away. Next slide.

Dr Marsha Treadwell:

So, across both sites, as Julie mentioned, we... Pain is so important in sickle cell disease. And the adult side actually has already a focus on implementing individualized pain plan. So, we will likely look to adding pain management itself as a quality indicator. We really need to include the voice of individuals with sickle cell disease participating in this APP supplement and find out what they think about the quality of an access to care.

Dr Marsha Treadwell:

We have an additional supplement to expand telehealth and that's being integrated with this APP supplement. We want to expand the sickle cell education for the APPs across the state, the region and the nation, and provide specific education about structural racism and its impact on sickle cell disease, as well as incorporate a focus on social determinants of health. Next slide. And I think that's it. Thank you so much.

Cynthia Powell:

Thank you very much, Dr. Treadwell and Dr. Kanter. We'll now have an opportunity for discussion questions and comments as usual we'll open it up to our Committee Members first followed by Organizational Representatives. And please remember to state your first and last names and make sure your mics are open. Let's see. Scott Shone.

Dr Scott Shone:

I didn't have my hand up, but I'm happy to make a comment.

Cynthia Powell:

Okay. I'm sorry. I thought I saw your hand.

Dr Scott Shone:

No. You were reading my mind Dr. Powell. I'll just... I just want to say real quick then while I have the mic that, great presentations and clearly, especially on the geneticists and genetic counselors' perspective, real clear need as we're moving forward with these disorders to expand that workforce. And I think it's interesting juxtapose next to some of the public comment on the need for the Committee to get ahead of new therapies and move expeditiously to add conditions to newborn screening, and further reinforces a constant drum beat that we've had at the Committee meetings over the past, at least years that I've been on the Committee across the different topics we've talked about, whether it's workforce education, continuity of operations, that this is a system and the programs alone that are being encouraged to expand lab testing and follow up, have to rely on a system that is dependent upon the workforce shortages we just heard about and so many other things.

Dr Scott Shone:

And so, I would encourage and look to partner with those who want to continue to expand at an exponential rate, that we work beyond just the newborn screening programs, but as a system to look at the genetic counselors, the geneticists and everything else that we need to make the system function to expand those as well, because, Dr. Kwon's presentation on neurology, just... There's so much that has to grow beyond just the tests and getting babies to care, to make this

work, to assure that we are finding and appropriately treating quickly. Thanks.

Cynthia Powell:

Mei Baker.

Mei Baker:

Sorry. Took a time to for me to unmute. Mei Baker, Committee Member. My question is for Dr. Treadwell. I wasn't thinking that in the future, because giving such a promising for gene therapy for sickle cell disease. So how does the training and education go hand-in-hand? Because I almost think our newborn screening, the education material, I have to start to think about this aspect because the equity accessibility, so could you make some comments on that?

Dr Marsha Treadwell:

Well, I actually defer first to Dr. Kanter and then I'll speak on it.

Julie Kanter:

All right. So, I think gene therapy is very promising, but I think we have a very long way to go before it's going to be universally available. I do think there's the potential for that, but we don't even know the optimal gene therapy. There are some significant barriers that we've recently come across. Hopefully we won't see again with the advent of leukemia as into sort of failed gene therapy, but I do think it's important in general, in newborn screening to impart so much more hope. I have many individuals living into their sixties and seventies with hydroxyurea and our newer therapies that are available, not just gene therapy, but crizanlizumab and oxbryta and other ones coming down the pipeline.

Julie Kanter:

And our increase use of exchange transfusions and then stem cell transplant itself. So, I think we have incredibly diverse new therapies and more are coming. So, I hope that helps answer, but I think gene therapy is going to be another five to 10 years before we get too far.

Dr Marsha Treadwell:

Yeah, exactly. And what I would just add is that in sickle cell disease, there has been limited focus on the disease modifying and curative therapies until recently. And the quality of life of people has really been neglected. And so, this increase in workforce, I think can address that while we were in the midst of beginning to roll out these disease modifying therapies and curative therapies. It's really important in terms of the quality of life and our APP who's the adult provider working on the pain management, she's... That's a huge focus. And again, it'll be a while before people are going to not be having those issues with pain. Thanks for that question.

Mei Baker:

Thank you. Yes.

Cynthia Powell:

Melissa Parisi.

Melissa Parisi:

Thank you. Melissa Parisi from NIH. My question is one that's a little bit more generic and I'm raising this for anyone to answer if they know. I think all of us would agree that one of the silver linings from this pandemic has been the increase in telehealth and telemedicine services being made available to families and Dr. Kwon, I was a little distressed to hear that reports are that states are now reverting back to previous restrictions on allowing cross state provisions of care for providers who may not be

licensed in a given state. I guess I'm wondering if given the, I would say overwhelming support for continuing in this mode, if at all possible, if there's any legislation, federal legislation or activities that any groups are pursuing that might actually try to establish standards nationwide for the provision of telehealth services.

Cynthia Powell:

Dr. Kwon, do you want to respond?

Jennifer Kwon:

I'm not aware of any, because it is complicated with the States. I think that the American Academy of Neurology and the Child Neurology Society, and probably any other society that has benefited from interstate telehealth would agree that we need some relaxation of these restrictions. But I think it's difficult for national entities to make these sort of state-based recommendations. But I will... Certainly it's one of my pieces of homework to go back to the CNS and AAS, to ask them what they're doing in this regard. Because we're definitely feeling it. I have families who drive across the border just to be in Wisconsin so that I can see them. So, we're doing a telehealth visit from their car basically, and that's better than nothing, but it does feel a little silly.

Cynthia Powell:

Cate, did you want to respond to that?

Cate Walsh Vockley:

Yeah. I just wanted to second what Dr. Kwon said, that really becomes an issue of states. I actually just yesterday got an email from the Pennsylvania licensing board saying that they were going to be discontinuing the waivers that were put in place when COVID impacted the system. So, it really is a state licensure issue. And I don't know how much influence the federal

legislation might have on something like that. So, we may have to go back to the old way of state by state.

Cynthia Powell:

Joan Scott, did you have a comment?

Joan Scott:

Yeah, thank you. I was just going to offer up potentially the services or information, HRSA runs the HRSA Office of Telehealth Resource Centers and there is one that does address national policy and issues, and we can certainly have them reported an upcoming meeting if the Advisory Committee is interested on trends and activities that are happening in this area.

Cynthia Powell:

Thank you. Okay. Robert Ostrander.

Robert Ostrander:

Yeah. Hi, Robert Ostrander, AAFP. I spend a lot of time in the adult world as well, and I think... What happens in the adult world can inform some of our discussions. I got into the sort of this whole universe 20-some years ago when my practice was part of a national learning collaborative on medical home for NICHQ for children and youth with special healthcare needs, and it was striking in that collaborative. About two third pediatricians and one third family physicians, how different in some ways, our approach to people with chronic conditions was because we have a lot of adults with complex chronic conditions. And I think there may be some lessons learned when we're trying to address some of these workforce issues in looking at how the adult primary care world deals with super complicated patients. And both through evolution and intent of a reasonably highly functioning primary care medical home.

Robert Ostrander:

A lot of us are pretty good at handling adults with very complex conditions. And I think maybe some of that model could be considered. It should look as to how to get workforce help from the primary care world as numbers of the medical home. One of the challenges though, and one of the differences is that a lot of our folks with complex conditions are reasonably common, so we have some experience, and we also have connection with our sub-specialists that we're working with. It becomes a little more muddy, whether it's adults or kids when it's a complex, rare disease. But I still think that's something that should be looked at when folks are exploring potential ways to stretch the workforce of subspecialists and geneticists for that matter.

Robert Ostrander:

I'm not at all surprised that they couldn't get interested with Project ECHO for PCPs, because I get several a year, I get an email or an invitation to join a six or 10 session Project ECHO for some specialized disease that everybody thinks primary doctors should know more about. So, I agree with that. I'm glad you tested that model, but I'm not surprised it didn't work. And I think it's good knowing that going forward. And to phrases this in form of a question, I just wonder across the group and especially maybe Dr. Kwon, partly because she and I worked together, but when she was back in western New York, had any thoughts about a model that involves the primary care physician in a way that allows some of the work to be shifted to the primary care physician without making the primary care physician the center of that specific disease's management?

Cynthia Powell:

Dr. Kwon, do you want to comment?

Jennifer Kwon:

Well, what I would say is that, I think that one of the things I thought was so interesting and inspiring from Dr. Kanter's and Treadwell's presentation was that it was a reminder of how disease specific these issues are. What a beautiful reminder of the lifelong chronic multi-system difficulties of caring for this particular disorder and how hard it has been and how challenging it has been to wrap our arms around it, to move it forward. And in many ways we have to recreate that for every one of these rare conditions that we face, some of them to a lesser degree that than others.

Jennifer Kwon:

And so, I would say that I am pretty bad at it in figuring out ways of involving primary care physicians on partly because the diseases that I sit in a silo with tend to be... They just tend to be more specialized. But I do see that changing. For example, I do think that for some conditions like X-linked adrenoleukodystrophy and late onset Pompe disease, I think their follow-up... I have seen how an engaged primary care physician can really make a big difference in terms of compliance and patient satisfaction and quality of life with those disorders. So, I have to think more about it myself.

Julie Kanter:

This is Julie Kanter, and I just wanted to tell you, we have worked with medical homes as well. It wasn't just isolated primary care physicians. The global STAMP project was everyone. If there were a primary care state provider in the state. And it wasn't just that they were invited to ECHO, they were actually given our phone numbers and email addresses. If they had a concern or problem or consultation for sickle cell disease, I was terrified we were going to get inundated. I can't tell you how disappointed I was to not get any calls. And I've also worked directly with medical homes, where the patients ended up actually

leaving the medical home in order to come into our center where they could get more specific care. However, what we have done and what I actually think has been incredible useful, and I did this in South Carolina and in Alabama, we actually have a primary care provider in my sickle cell center.

Julie Kanter:

And that's what has been done in many other places as well, Ohio State, Atrium health, just to name a few. And we have seen incredible satisfaction from patients. We're actually now hiring a psychologist as well, because patients have lives too. And so, they don't want to have to go to their sickle cell specialist and their primary care doctor and their psychologist when they're trying to work and maintain a family. So, we call it the one-stop shopping model. And it's not going to be feasible for everyone, but we've... The amount of increase in primary care complications and needs addressed has just tripled. So, I think that the medical home model is fantastic, but I like it as a specialty medical home model in some of these very complicated patients.

Cynthia Powell:

Jed Miller.

Jed Miller:

Yes. Hi, Jen Miller, Association of Maternal And Child Health Programs. I was going to say that I'm going to ask the converse of what Dr. Ostrander was asking, but then Dr. Kanter, what you said is going in this direction is, and I'm curious about panelists experience where, because of family preferences, they desire the specialist to be the medical home.

Jed Miller:

I know you just mentioned about Dr. Kanter about subspecialist medical home. I'm wondering especially

for Dr. Blitzer, but not only for you with respect to the uncombined and residency in pediatrics and medical genetics, I'm not sure if you capture any data about how providers practices actually look. In that scenario, they'd be armed to be, they could provide well-child visits as well as do the specialty care.

Jed Miller:

I'm just curious if there's any experiences on that front in terms of functioning as a medical home with or without the routine health maintenance measures such as immunizations. On somewhat related note, Dr. Kwon, you were talking about seeing children longer than you might otherwise into early adulthood. I'm wondering if there's any experiences with that life course and actually caring across generations.

Jed Miller:

Meaning, if you see children long enough, perhaps they will become parents to themselves. Just thinking about even parenthood itself could be thought of as a long-term follow-up measure of sorts, outcome in certain ways. Just curious if any panelists have any comments on those topics.

Cynthia Powell:

Mimi, do you want to take that first?

Miriam Blitzer:

I'll start briefly. Thank you, Dr. Miller. From the standpoint of individuals who are certified or trained and then are ultimately certified in two specialties and let's concentrate the majority of those would be peds and medical, genetics, and genomics. We do and I'm going to let Dr. Powell comment afterwards as this is her area as well. We do see since geneticists do see patients across the ages.

Miriam Blitzer:

There's not the pediatric restriction on them that metabolic patients many times continue with their geneticist, who may also be a pediatrician, but really as a geneticists, managing their care. At many situations, at many institutions, it does become their primary care home, their medical home, to some extent. The board in and of itself does not do practice surveys, our college does.

Miriam Blitzer:

Also, in the workforce study, we did get that feedback as well. I would say in general, we do see people going into the combined and those that are interested in metabolic, certainly managing patients as they transition from pediatrics to adulthood in their own families. I wanted to also mention that we have this relatively new specialty. The first exam was given in 2009 of a subspecialty in medical biochemical genetics. It is a group that is growing.

Miriam Blitzer:

Again, the numbers are small because it's a new specialty or subspecialty, but it's clearly the area of growth where most individuals who are completing or many of the individuals are completing their residency and training in peds, in medical genetics, or in the combined are also training in the subspecialty within almost doubling from 2017 to 2019 in the number of individuals certified and moving forward. I do think the answer is that the medical home would be there for those groups. Dr. Powell?

Cynthia Powell:

Yeah, I agree. I think we are training a number of individuals now in who have a background in internal medicine, which has really been very helpful to help as our patients with rare disorders transition to adulthood. It's been extremely difficult to find

providers willing to take care of them in any area, even specialty care. I'll turn it over to Dr. Kwon.

Jennifer Kwon:

Thanks. I think this is where it's helpful to remember that I work as part of a team. In many places, in many medical centers, it is difficult for a primarily pediatric provider to see patients easily into their adult years, emergency rooms. I think it requires me to become familiar with the adult medical care system, which can be challenging for a variety of reasons.

Jennifer Kwon:

As soon as my patients get to that 18-to-21-year transition period, I will say that at this point, I tend to try to transition them to adult neuromuscular providers, for all kinds of practical reasons. Even the nurse practitioners that many of us work with may only be licensed to take care of patients who are pediatric patients. I think that there's this ripple effect, like who will the emergency room see?

Jennifer Kwon:

Where will they be admitted? Having the adult age makes it challenging. Sometimes, I'm not sure what a medical home is. I feel like that's a term that's used less in Wisconsin than it was in New York when I lived there. I think that patients who have rare disorders identified by newborn screening, who have chronic diseases, that require a lifetime of care, need a medical network.

Jennifer Kwon:

They need a medical support system that we have to help identify and help maintain through their life cycle. I will say that for example, with SMA, I follow a number of patients into their adult years, and it is fascinating how many of them want to be parents, and are making decisions about treatment based on that. I

do think that the system is unfortunately feels very fragmented and broken right now.

Jennifer Kwon:

It doesn't make it easy for us to create those lifetime networks for patients with chronic diseases.

Cynthia Powell:

In order of those who I've seen with their hands raised first, we'll take Natasha, then Siobhan, then Georgianne and Debra. You'll be after Georgianne. Natasha?

Natasha Bonhomme:

Great. Thank you so much. Natasha Bonhomme, Genetic Alliance, I have a two-part question mostly for our first two speakers. I wonder if you could speak a bit to what the vision is for the growth of the workforce in genetics. Are you looking at hoping for 15% to 20% above what we currently have? Is it measuring the length of time that someone has to wait to go and see a genetics service provider and reducing that to X number of days, just trying to get a better sense of what would success look like?

Natasha Bonhomme:

Not necessarily just for today, but where we're projected to go, knowing that there are projections around gene therapy, and all other parts of the system. Then also, my part two or second question is it was really interesting to see the growth of genetic counseling programs, as well as the growth of student enrollment.

Natasha Bonhomme:

Just wanting to get a better sense of is the growth in diversity equal to that more or less, just putting that into context knowing that particularly in the past year, there's been a number of different work groups

and initiatives really looking at that issue? Thank you.

Cynthia Powell:

Mimi?

Miriam Blitzer:

Thank you, Natasha. Those are excellent questions and topics that we're thinking about very seriously. The question of growth, and that's what we're looking at now is first understanding what has the growth been and do we see growth? Then also, understanding how many clinical geneticists' laboratory and other of the workforce working with genetic counselors and dieticians and other providers, nurse practitioners do we need?

Miriam Blitzer:

From the standpoint of ABMGG, and we really are looking with the college on this, since we are the certifying arm, is that we would really hope that we begin to see. My personal thoughts would be a significant increase of 10% to 15%. Looking at it consistently, the CS slope begin to go up. To that end, I wanted to touch base on your second topic.

Miriam Blitzer:

We are a relatively new specialty, although it sounds like we're pretty old at 30 or 40 years, but we are relatively new, to recruit into our specialty. Let me first talk about, let me just only talk about clinical geneticists recruiting from med school. We have to be on the forefront. When medical students get into their third or fourth year of training, there isn't a requirement that, "Do a medical genetics rotation." For instance.

Miriam Blitzer:

Sometimes the goal is how do we get them as excited and recognize what this field is all about and what the promises are? I do think the combined residencies have helped. I do think that a better reimbursement for clinical geneticists will help. We're not there. From the standpoint of diversity, that's a huge issue, not as in our specialty, but it's one that we're focusing on.

Miriam Blitzer:

One of the things we're doing is definitely, as I mentioned before, right now, we're in the midst of trying to look at what's the pipeline look like compared to clinical geneticists and medical geneticists that are already in practice. We are actually a relatively young specialty as far as looking at the age of our diplomas, but we need to look at this. We have a task force working on this.

Miriam Blitzer:

One of the things is that we still have open slots, open positions for residents in certain programs. One opportunity there is how do we, not only attract medical students, but underrepresented minority medical students into our specialty, and have them recognize the field. For instance, out of the four HBCU medical schools there's not one clinic board certified clinical geneticists at any of those four.

Miriam Blitzer:

There's not really good role modeling or exposure. How do we partner and do this to that end? I'm working with the Dean at one of the HBCUs on how to increase exposure and excitement to our field. Recognizing how important it is that they be an incredibly competitive group to come into our field and we need them. I'm going to stop there. Thank you.

Cynthia Powell:

Siobhan Dolan.

Siobhan Dolan:

Good afternoon. Thank you so much for this extremely interesting set of discussions this afternoon. My question is for Cate Walsh Vockley. Cate, I was wondering if you have any thoughts or comments on the role of genetic counseling assistant as a way to flesh out and expand the genetic counseling? That's actually the question. Do you see a role?

Siobhan Dolan:

Where does that work? How could that fit in a genetic counseling assistant role around this newborn screening and increasing genetic counseling capacity?

Cate Walsh Vockley:

We have been using genetic counselors in general genetics clinic, not so much in the newborn screening setting at this point, although I do believe that that is one way that we could increase capacity with having. Here in Pittsburgh, our genetic counselors are really helpful with some of the coordination of appointments with coordination of testing, reaching out to family members to get samples.

Cate Walsh Vockley:

A lot of the nuts and bolts of getting things done. I do think that that is one area where we have been able to utilize non-genetic counseling trained individuals. They do have to go through a period of training themselves, but oftentimes it's within the clinic setting that they're trained. I know some of the programs do have a one-year genetic counseling assistant training program as well.

Cate Walsh Vockley:

I do think that's been a big help and could be applied in newborn screening as well. Relative to the previous

comments in terms of growth of the profession, genetic counseling training doesn't exist in a vacuum. We are bound by available clinical training sites and the requirements of the accreditation, and board in terms of moving forward with more programs.

Cate Walsh Vockley:

I think the anticipation is that we will continue to grow. I think the growth rate has been so significant in the last five years that it will be hard to keep up that growth rate. I think there are a lot of people interested in doing that. As far as the diversity question, I know, looking at our programs incoming students, there's been a tremendous effort to increase diversity in the programs.

Cate Walsh Vockley:

The program directors organization has been making efforts in that regard. I do think that our program, at least a third of the students are considered underrepresented, maybe closer to a half. I think it's doable. I think we can reach out to the communities and increase diversity in our field.

Siobhan Dolan:

Thank you.

Cynthia Powell:

Shawn McCandless. Did you?

Shawn McCandless:

Thank you. This conversation is both enlightening and very frustrating. I've been just writing down lists of notes of things to discuss. I have a lot of questions about number one, what is the role of this committee in thinking about this issue? I understand this is more for informational and brainstorming, but these workforce issues are incredibly complex.

Shawn McCandless:

Some of the thoughts that I've had are, first off, is there really a shortage? Are there shortages and who determines the workforce need? I think if you look at the NSGC website, they have the National Society for Genetic Counselor states explicitly that there was not a shortage of genetic counselors. They have pretty good information to support that on the website for physicians.

Shawn McCandless:

With that said, there's clearly a deficiency in certain areas in the U.S., and the same is true for positions. There are genetics clinics for physicians in the U.S. where you can be seen with very little weight. There are other places where you have to drive hundreds of miles and still have a six to 12 month wait to be seen. It's a shortage maybe, but it's also a distribution issue.

Shawn McCandless:

All of the problems that have been addressed with diversity is there are problems with salaries. There are problems with getting providers where they're needed. All of this, in my opinion, comes down to the fact that we don't have a healthcare system in the United States. We have basically a healthcare free market, a free-for-all, where as if you can get somebody to pay for something, you will do it. It's a totally messed up marketplace.

Shawn McCandless:

None of the things that we're discussing are going to be solvable in my opinion, in the absence of some meaningful reform of our healthcare system. That brings me back to where I started, which is, what can this committee do? What should we be doing? I think that the folks who were talking about sickle cell and the development program for ADPs had the right target, is

that we should be encouraging ways to really specifically identify what is missing in our newborn screening systems and follow up.

Shawn McCandless:

Then, figuring out creative ways to address those specific needs. I'm not sure that we're getting there and I'm not sure what the mechanism for getting there is. I'd be curious what others have to say.

Cynthia Powell:

Georgianne Arnold?

Georgianne Arnold:

Well, Siobhan stole a little bit of my thunder, but the SMID has recently convened an ad hoc committee looking at workforce. I participated in a few projects with Mark Corson from Rare New England on workforce recruitment. One of the significant concerns of the candidate to come to the recruiting sessions is money, not because they want to be rich doctors.

Georgianne Arnold:

If you want to be rich today, you become a hedge fund manager, but because they have $200,000 in loans and they can't really go into one of the lowest paying medical specialties. With this group, having a theoretical federal connection, I'm wondering if you look at the NIH loan repayment program for people doing research, could newborn screening be added to that, so that there's some loan repayment for people who work within the newborn screening field?

Georgianne Arnold:

Could there be a small grant program for recruitment seminars, that we could go out in and literally get pizza and Coke and go to medical students, and college students and try to recruit them, educate them, and recruit them into the field? I would really like to see

us perhaps utilize available resources to help ease the burden of going into this field.

Georgianne Arnold:

After talking to some college and med students fairly recently, it's real. The worry that they won't be able to afford a house or send their kids to college because they've gone into medical genetics is real.

Cynthia Powell:

Debra Freedenberg.

Debra Freedenberg:

Some of the questions I have other people have raised, especially even in a state that appears to have a fair number of clinical geneticists that doesn't really reflect what's happening across the state. There may be a large number of geneticists concentrated in one area of the state, while other areas who have significant population centers are underserved for genetics and may have no access to genetic services.

Debra Freedenberg:

There are large underserved areas where there may be a concentration of geneticists in one large urban area. I think that's true for a lot of other States as well. Another issue was this transition issue from pediatrics to adult and medical homes. We dealt with a fair number of folks who've undergone that transition, and it's always a difficult period. The feedback that we've gotten is that pediatric medical homes tend to have wraparound services.

Debra Freedenberg:

When those patients wind up in adult medical homes, often they don't find those same wraparound services, and it's been rather bumpy transitions as well for them. Then, the third point that I just wanted to make is a lot of clinical geneticists aren't pediatric-

based. If you're based in a children's hospital, you may have a hard age cutoff that pediatric hospital may say, "You are not seeing anybody over 18 or over 21, or whatever it is."

Debra Freedenberg:

Dr. Kwon has alluded to that. It's a liability. We're not used to handling somebody with a heart attack or whatever. I know, I myself in was pulled into my chair's office one day when I was treating an adult with Pompe Disease as to why I was treating an adult when I was based in a children's hospital. I think that's a very real issue that's out there, and that will continue to be out there in terms of geneticists who are based in pediatric centers are not viewed as being able to treat adults, even though most clinical geneticists do treat adults.

Debra Freedenberg:

I think that you need to recognize depending on the setting where they are, that it's also a barrier to continuing care for long-term care as well. In terms of recruitment and involvement of geneticists in newborn screening programs, it can be very difficult to integrate in a system that is not used to a specific category of genetic counselors to integrate them into a state program.

Debra Freedenberg:

It's not impossible, but it is a large task. I think almost everything else someone else has brought up. Thank you.

Cynthia Powell:

Thank you. Scott Shone, you had your hand raised. If you want to say something, I'll give you the final word.

Dr Scott Shone:

I think what Georgianne mentioned around more incentive-based opportunities is where I think we need to be thinking, and I would expand it to the whole public health workforce, not just newborn screening. I think across the board, we've learned that there are challenges getting people to come into public health. The administration announced some huge opportunities yesterday to reinvest in newborn screening.

Dr Scott Shone:

As we've discussed, I think what's important for those of us who have broader roles in public health beyond just the newborn screening system as these opportunities come about because of the pandemic. We make sure that all of the pieces that newborn screening touches are also incorporated in all of these long-term infrastructure changes that we're making to the public health system.

Dr Scott Shone:

Again, I just, I hate to sound like a broken record, but I keep coming back to system and we all need to be aware of how integrated we are. When we make these adjustments, we're thinking about all the upstream and downstream activities and people and processes from where we stand and look at that. I appreciate Georgianne's comments around that. I didn't become a public health laboratory director to be wealthy.

Dr Scott Shone:

I certainly wouldn't be as fulfilled if I were a hedge fund manager. Luckily, I have a spouse who understands and understands that. Anything we can do to help make these roles that we are so passionate about more attractive. For our teams, especially, who always solve every problem is critical. Thanks for the opportunity, Dr. Powell.

NEW BUSINESS

Cynthia Powell:

Thank you. Thanks once again, to all of our presenters for this session. Certainly, an area that many of us are extremely interested in and hope to have further discussions in the future about. Since we are running a bit over, I wanted to ask if the Committee members have any new business or announcements? We don't have a lot of time for discussion. However, thinking about things for future discussions, future meetings will be helpful and welcome. (silence) Melissa Parisi.

Melissa Parisi:

Hi, this is Melissa Parisi from NIH. I just wanted to announce an upcoming three-day workshops spread over three weeks, three Thursdays in June. It is being sponsored by NIH and led by the National Center for Advancing, NCAT, I cannot remember what it stands for, but essentially the title of the workshop is Gene-targeted Therapies Early Diagnosis and Equitable Delivery.

Melissa Parisi:

This will be Thursday, June 3rd. Thursday, June 10th and Thursday, June 17th. I don't know if there's anything that comes out of this. I'm happy to post the registration page for anyone who might be interested.

Cynthia Powell:

Thank you. Shawn McCandless.

Shawn McCandless:

Thanks, Cindy. I had emailed Cindy and Mimi after we had the public comment earlier this morning from the EveryLife Foundation and from the Society of Gene & Cell Therapy. I really think that we need, I know that there's been a work group and we have been working on a

plan for the evidence review and the evaluation of new disorders.

Shawn McCandless:

I think it feels to me like, the points that those public statements made today was to warn us that there's a crisis coming in terms of new nominations and our ability to respond to them. I think what we've seen from the MPS II process and the most recent processes is that we're going to be really challenged to address 10, 20, 40 new nominations over the next few years with our current system.

Shawn McCandless:

I just really hope that we can spend some time at our next meeting, identifying specifically what the issues are that need to be addressed. Then really, just trying to be creative about fixing the system, or fine-tuning the system to make it more responsive and be able to turn things around faster. I think this has to be the 800-pound gorilla in the room that this Committee has to face, because it is one of our, if not the primary function of the Committee, it seems to me to make these.

Shawn McCandless:

To advise the Secretary of HHS about the ruts. I just want to put it out there that I think we really need to focus on that very soon.

Cynthia Powell:

Thank you, Shawn. Anyone else? Our next meeting via webinar will be August 12th through 13th, and this meeting is now adjourned. Thank you all.

Shawn McCandless:

Thanks, Cindy. Thanks, everyone.

Jennifer Kwon:

Thanks, Dr. Powell. Thank you.

ADJOURN