THE ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN

IN-PERSON/WEBINAR

HRSA HEADQUARTERS 5600 FISHERS LANE

ROCKVILLE, MARYLAND 20852 (Pavilion)

Friday, August 11, 2023

10:00 a.m.

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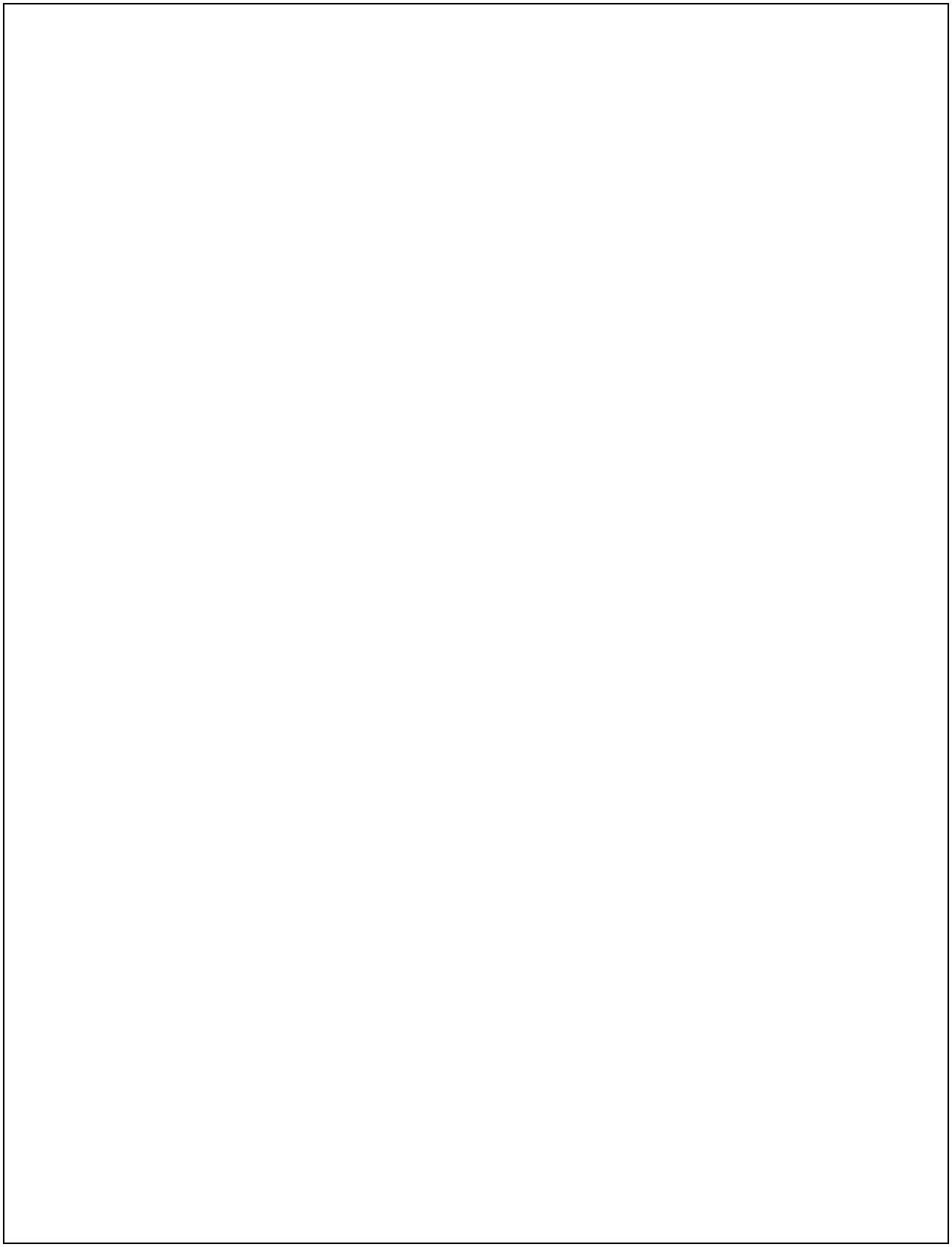

P R O C E E D I N G S

10:00 a.m.

Welcome and Open

DR. CALONGE: Good morning. I want to welcome everyone back to day 2 of the ACHDNC August 2023 meeting. Before we do roll call, I want to reintroduce an individual that many of you know very well. Cindy Powell is serving as the new organizational representative for the American College of Medical Genetics and Genomics.

You probably remember that Cindy is a Professor of Pediatrics and Genetics at the University of North Carolina at Chapel Hill, at their School of Medicine. She's a board-certified clinical geneticist, cytogeneticist, pediatrician, and genetic counselor, and is the immediate past Chair of the ACHDNC, and a member of the Board of Directors of the American College of Medical Genetics and Genomics.

So welcome back, Dr. Powell, you didn't stay away very long, and we're happy to have you. I don't know if you would like to make a comment or two?

DR. POWELL: Hello. Hi everyone.

DR. CALONGE: Thanks for rejoining us and continuing to support the work of the Committee. That's super.

DR. POWELL: Nice to be back and see a lot of familiar faces. Thank you very much.

DR. CALONGE: So with Cindy being back, we're going to just try to move forward. Today we're going to begin with a presentation from Dr. Houtrow on Focusing on Equity After Newborn Screening. We'll then have public comments, and after that a break.

We'd like to just quickly review the documents that I sent out last night trying to capture the additional information and ideas around expedited review, and try to provide you with two formats, the narrative document, and the slides with the evidence we discussed.

And then we'll move on hopefully, to talk about Krabbe and whether or not you would like to move it on in the expedited review process. And I'm going to turn things over to Leticia, who will do roll call, and then Dr. Kemper will -- I'm sorry, Dr. Houtrow will begin her presentation.

COMMANDER MANNING: Great. Thank you, Ned. From the Agency for Healthcare Research and Quality, Kamila Mistry?

DR. MISTRY: I'm here. Thank you.

COMMANDER MANNING: Michele Caggana?

DR. CAGGANA: Good morning, here.

COMMANDER MANNING: Cynthia Hinton from the Centers for Disease Control and Prevention?

DR. HINTON: Here.

COMMANDER MANNING: Jannine Cody?

DR. CODY: Here.

COMMANDER MANNING: From the Food and Drug Administration, Paula Caposino? From the Health Resources and Services Administration, Jeff Brosco?

DR. BROSCO: Here.

COMMANDER MANNING: Jennifer Kwon?

DR. KWON: Here.

COMMANDER MANNING: Ash Lal?

DR. LAL: Here.

COMMANDER MANNING: Shawn McCandless?

DR. MCCANDLESS: Here.

COMMANDER MANNING: The National Institute of Health, Melissa Parisi?

DR. PARISI: Here.

COMMANDER MANNING: And Chanika Phornphutkul?

DR. PHORNPHUTKUL: Here.

COMMANDER MANNING: And for our organizational representatives, the American Academy of Family Physicians, Robert Ostrander?

DR. OSTRANDER: Here.

COMMANDER MANNING: From the American Academy

of Pediatrics, Karin Downs?

MS. DOWNS: This is Karin. I'm from AMCHP.

COMMANDER MANNING: AMCHP, my apologies.

MS. DOWNS: No worries.

DR. FREEDENBERG: And this is Debbie Freedenberg, and I'm from AAP and I am here.

COMMANDER MANNING: Yes. Got it. Thank you for the correction. From the American College of Medical Genetics and Genomics, Cynthia Powell?

DR. POWELL: Here.

COMMANDER MANNING: From the American College of Obstetricians and Gynecologists, Steven Ralston? From the Association of Maternal and Child Health, Karin Downs, thank you. From the Association of Public Health Laboratories, Susan Tanksley.

DR. TANKSLEY: Here.

COMMANDER MANNING: From the Association of State and Territorial Health Offices, Scott Shone?

DR. SHONE: Here.

COMMANDER MANNING: From the Association of Women's Health Obstetric and Neonatal Nurses, Shakira Henderson? From the Child Neurology Society, Margie Ream?

DR. REAM: Here.

COMMANDER MANNING: Department of Defense,

Jacob Hogue?

DR. HOGUE: Here.

COMMANDER MANNING: From the Genetic Alliance, Natasha Bonhomme?

MS. BONHOMME: Here.

COMMANDER MANNING: From the March of Dimes, Siobhan Dolan?

DR. DOLAN: Here.

COMMANDER MANNING: From the National Society of Genetic Counselors, Erica Wright?

DR. WRIGHT: Here.

COMMANDER MANNING: And from the Society for Inherited Metabolic Disorders, Susan Berry? Okay. And I am just briefly going to go over, remind folks about conflict of interest. Please note that you must recuse yourself from participation in all particular matters likely to affect the financial interests of any organization with which you serve as an officer, director, trustee, or general partner, unless you are also an employee of the organization, or unless you have received a waiver from HHS authorizing you to participate.

As in the case today when a vote is scheduled, or an activity is proposed, and you have a question about potential conflict of interest please notify me

immediately. You can email me at lmanning, so L-M-A-N-N-I-N-G @HRSA.gov. Thank you, and I turn it back over.

DR. DORLEY: This is Christine Dorley. I didn't hear my name called.

COMMANDER MANNING: Yes. Thank you.

DR. DORLEY: But I'm present, thank you.

DR. CALONGE: Thanks everyone. I did, as I was listening to the roll call, I just wanted to pause and give a shoutout to our organizational representatives. Your attendance and participation is so greatly appreciated. We know that we made you sit for long times on Zoom calls, or in meeting rooms, but it's so key to have this two-way communication with the organizations, and viewpoints that you represent, so thanks for attending and thanks for being here for day two.

Focusing on Equity After Newborn Screening

DR. CALONGE: Moving on, it's my pleasure to introduce Dr. Amy Houtrow. Dr. Houtrow is a Professor and Endowed Chair in the Department of Physical Medicine and Rehabilitation for Pediatric Rehabilitation Medicine at the University of Pittsburgh School of Medicine. She is also the Vice Chair for Quality and Outcomes, and is the Chief of Pediatric Rehabilitation Medicine Services at UPMC Children's Hospital of Pittsburgh.

Dr. Houtrow's main clinical focus is caring for children with disabling conditions, and helping to improve function and quality of life to the greatest degree possible. Her patients include children with spina bifida, cerebral palsy, hematologic disorders, brain and spinal cord injuries, and orthopedic musculoskeletal and neurological disorders and conditions.

Complementing her clinical focus, Dr. Houtrow's research focus is on childhood disability trends, advancing health equity, and developing channels to improve service delivery. I'd like to turn things over to you, Amy, and thanks so much for joining us.

DR. HOUTROW: Well, it is a distinct pleasure for me to be here. And just for everyone, I'm going to do a visual introduction as well, in case we have anyone on the call with vision impairment. I'm a pale woman with shoulder length dark brown hair. I'm wearing a bright yellow shirt today, and I have black glasses on, and there's pictures of animals on the wall behind me. I get the distinct pleasure of speaking to you all about the issue of equity as it relates to what happens after newborn screening.

I will start off, though, with a couple of disclosures. I have quite a lot of grant funding, none

of it relates to heritable disorders and so no actual conflict. I'm going to speak to you about thinking about equity, so to do that I want to just make sure everyone's kind of on a level playing field with some definitions.

I know that many of you know these inside and out, but if they're new to other people I think it's really helpful for us to just level set. And I'm going to review some issues around access to care, and we're going to pay special attention to children who come from minoritized, or otherwise oppressed backgrounds.

And all the while I want us to be thinking about in our own work how do we engage in the work we do without the equity lens. So this is just stage setting because all of us in the work that we do have growth potential, myself included, around how we approach and design and develop programs and services in such a way that it optimizes health equity.

So health equity, that just means that everyone has a fair and just opportunity to be as healthy as possible, so fair and just. And it's focused on this opportunity. And it's very process based, because it requires us removing obstacles, that is obstacles such as the root causes of health disparities, poverty and discrimination, and then all of their downstream

consequences. And these are really about lack of access to the things people need.

So when we think about health disparities in the lens of health equity, we recognize this as differences in health, or its key determinants that adversely affect those that are marginalized or oppressed. And the process forward to that health equity is the actual reduction in those disparities.

So I love a good picture, and so I put this into an upstream downstream graphic for you. So I just mentioned poverty and discrimination, and all of the kind of political, and I'm using the word political because there's policies that drive -- I'm not using the word partisan, our access to things like housing, finance, employment, and those things.

And all of those are amenable to policy interventions because what we recognize is there's a lot of issues here. Housing and food instability and insecurity, lack of stable employment, lack of good education. And then in the world that I swim in, right, the lack of access to services and supports that promote health.

So if we have this baby who just had their newborn screening, we don't want them out here in the open water. We want this baby to get all the support it

needs to travel the journey through life as healthy as possible. So you heard me say the word minoritized before, and I think it's just important that we own how deeply structured racism is in our culture, society and policies.

And so structural racism is the totality of ways that society fosters discrimination, or oppression. It's all those different things that I just said. So systems that limit access to housing, good education, employment, earnings, all of those things are mutually reinforcing.

I know everyone is really familiar with terms around racism, but I'm guessing people know basically about classism, but not really so much how it operates. And it operates in conjunction oftentimes with racism and other sources of oppression. So, it is the systematic oppression, people who are disadvantaged based on their class or financial worth.

And what happens here is that the policies and the systems and practices are repeatedly in this circular way to benefit the upper classes by keeping or oppressing people below them. It is, of course, held in place by a system of beliefs and cultural attitudes that really see people who are poor as less worthy. And we have this all over the U.S. Ideas like, well, in

America all you've got to do is lift yourself up by your bootstraps, right?

And you can't actually lift yourself up by bootstraps. That's the whole point of the phrase. And so the ridiculousness of it is that if you just work hard in America, you can be the next millionaire, when all of these kind of structures are set up to not allow that to occur.

And this is really important because kids often live in poverty. So what I'm showing you here is over the last 20 years, and this is when the great recession happened, so I grayed this out. We saw a massive increase in childhood poverty, and this line here is the "ALL" children. So I'm showing you this by race.

And what's really concerning here is there's a big differential, so white children are less likely to be poor, as are Asian children and Hispanic children, and black children. Okay, this is not just a little difference, so all children, nearly 10 percent to 27.7 percent of black children. So we're talking about essentially three times the rate of poverty.

So I told you that poverty, classism and racism often go together. They also often go together with ableism. So ableism is that same sort of thing that we've been talking about. The stereotyping, the

prejudice discrimination, oppression, but this time not by race, or not by income, it's towards people with disabilities where people with disabilities are thought of as a diminished state of being.

So whether it's the ideal physical state, or a cognitive state, people with disabilities are deemed less worthy. And I put up the ICF because I really love how this neutralizes language. But what I can tell you is like all that stuff when I was showing you the upstream and downstream, the international classification considers them environmental factors, and of course they interact with our own personal choices and beliefs, and desires.

But it's really important to recognize how incredibly essential it is to understand the environmental contexts, so those social and political determinants. So why am I telling this to people? Babies who show up with a positive newborn screening, you know, are heading down the path to being children, youth with special healthcare needs, so they are more likely to be poor, and they are more likely to be minoritized.

So this is just an income breakdown, so these kids are living below the federal poverty level. These kids are -- their families are quite well off, above 400

percent. And what you can see is just this big, huge number here, minoritized black families, and Native Hawaiian and Pacific Islanders, so many more of those kids with special healthcare needs are living in families that are poor.

And I'm certainly not an expert in newborn screening, but I do see a lot of kids in my practice who have the disability consequences of sickle cell disease. And you know, this means these kids have been kind of undermanaged for a long time, that they were ending up with these adverse disabling consequences, such as related multiple, repeated strokes.

So we know, obviously, that sickle cell disease has a strong hereditary component based on like racial grouping, so shared and ancestry. So we know these kids, these African American kids, just are more likely to live in poverty, and they're more likely to experience the consequences of racism.

So I think it's really relevant as we think about our successes in identifying a disease versus our successes in helping children thrive and have optimal health when that disease is identified. And of course, we know that there's a lot to be done to improve care for individuals who have sickle cell disease, and certainly I see that in my own clinical practice.

I also see a lot of kids who have failed newborn hearing screen, but also get lost in the shuffle of follow-up. And you know there's been, and I pulled this from the Maternal and Child Health Bureau website, there's been a lot of achievements here.

So really, we were doing terribly in terms of getting kids screened. Now basically all the kids are being screened, so this is a big, big win. So like 97 percent of infants. That is a hip, hip, hooray. But what is not a hip, hip hooray is that there's a hefty portion of them that are just not getting the evaluations and follow-up that they need.

And what do we need to do for kids who have identified hearing impairment is we need to get them engaged in services, and we know the earlier you intervene the better things are. And there's this time window from when children, infants leave the hospital and they have this like just robust opportunity for us to intervene on their behalf, and so often that intervention does not occur.

And guess who that intervention does not occur for most? The children who come from oppressed and minoritized backgrounds. And these are the kids that really need to get access to high-quality healthcare, but just are not in the same way. So I'm going to share

a bunch of datapoints, and these are all from the National Survey of Children's Health. And you can actually go to ChildHealthData.org and run these same kind of different analyses.

So, as you guys know, the Maternal and Child Health Bureau has identified systems of care, six activities that identify that kids with special healthcare needs are getting care in a well-functioning system. So, first of all, I just have to say overall our ability to do this for kids is crap-tastic.

So this is like not even 14 percent of kids with special healthcare needs get it. But you know the ones who do? Are the kids who are white; and the kids who don't -- are from the minoritized backgrounds. So even though we're not doing well overall, we are doing less well. So this is one of those opportunities for health equity.

This is where we try to think about what things would make it more likely that these kids, these oppressed kids, could get what they need from the system. So again, I'm going to show you, this time by family income. The lightest green here are the kids in families who have the least amount of money.

The darkest green is the families who have the most amount of money. And again, overall, we're just

not doing so well. That's the same line that I showed you on the previous slide. But of course, who is doing better? Kids who come from more well-off families. Who is doing worse? These kids who are living in poverty or near poverty.

And you know I couldn't even create a better stepwise relationship between income and care in a well-functioning system than the one that already shows up in the data, but just tells us we are so desperately needing to do something that helps the kids that need the things the most get the things that they need.

So when I think about the kids that I cared for, and many of the children who come from the world of newborn screening, is the kids who have more complex healthcare needs. So in this graphic I have kids who are less medically complex. Their healthcare needs are relatively easy to manage. And these kids are the ones who are more complex.

And of course, that same line would be here of the 14 percent, less than 14 percent, because that's the baseline line for care in a well-functioning system. And I want to rephrase this into this is opportunity. So nearly over 80 percent of kids who have less complex healthcare needs have an opportunity to have better care in a well-functioning system.

And the opportunity is even bigger for the families of kids who are more complex to be able to get the care for their child that they need. And some of the core things are just like how we engage with families, and so I just wanted to share family-centered care by income. A kid with another stepwise relationship. So basically family-centered care is that you feel like a partner. Your providers respect your values and understand your goals, that you're not in this kind of like talk down. You're engaged in discussions about what happens next, and what you want for your child.

And well-off families tell us they get that, and very luckily over 85 percent of families report this overall. But, again, this ridiculous stepwise relationship where the kids we need to provide the most services to in the best way are just not -- their families are not reporting that service.

And also, by minoritization, again, we look pretty good overall, but it's the white families who report it, or marginalized families who do not, which really says that there's an opportunity here. And one of the hardest things is moving from a screening to a full evaluation and treatment. We have a lot, a lot of drop-off, so not just in the newborn screening space,

but also in the, you know, screening for developmental concerns, and the primary care pediatrician's office, for example.

And one of the things that families repeatedly tell us, and it's illustrated in the National Survey of Children's Health, is how frustrated families are at getting care. So I have combined the more complex, less complex, kids with special healthcare needs with the income, and with racial differences, by minoritization. Well, you know, for the kids who have the more complex conditions, right, many of these kids would show up with newborn screening identified by conditions on their screening, their families are reporting they have frustration getting care.

So I work with kids who have neuromuscular disorders, and you know, these are things that we genetically know about, and yet and although we have systems that are set up, multi-disciplinary programs still, you know, families whose kids are more complex are just reporting they're so frustrated getting what they need.

And also, it's real consequences for their family's functioning. So families also report that they have to cut back on work to just meet the needs of their child, so more families who are poor are reporting that

they have to cut back work than families who are well off.

Okay, so who needs the income the most? The families with the least income. Right? These are the things that just drive me totally bananas and upset, because these are the things that I recognize that we need to address to make sure that families are functioning well, kids are getting what they need.

I don't even need to show you the next slide. I will, and talk about what it looks like by minoritized race in terms of having to stop and cut back work, but you know who it is? It's our minoritized families who are doing that.

So just the kind of service, if you think about it really robustly, these families are not getting what they need, and they often report to us that they have trouble paying the medical bills. If you cannot pay your bills, you often do not seek care.

So again, I've kind of layered these all into one slide. So these are the colors you're familiar with around income. These are the minoritized groups, and then this is by complexity. So families who have the more complex conditions, heritable disorders like muscular dystrophies, or sickle cell disease are really reporting trouble paying their medical bills. It also

really relates to insurance as well.

But I want to tell you a couple of stories because I think this is some of the things where we put on our health equity eyeglass lenses, right? And we start to see how things are set up and are operating. And it feels like we have opportunities to shift, right? Like what are the unintended consequences that end up hurting people who are already oppressed, limiting their options?

So I'm going to give you an example of attendance policies, so let's say you're a parent of a child, and you need non-emergency medical transportation for your child to get to their therapies, or to their evaluations, or to the doctor, okay. So non-emergency medical transport is great, but it's super challenging to use.

So it provides you a two-hour window usually for pick up. Two hours, right? That's not how I plan my days with this like big lag time of two hours. Also, by the way, it's frequently late, and occasionally they cancel at like the very last minute where you wouldn't even have time to make other arrangements.

So what happens is that the providers on the other side are like oh my gosh, another no show. Oh, you didn't even call us to tell us they couldn't come,

like they don't. And then we get into all of these ridiculous thoughts. They must not really care about their child's health. I mean, they must be, oh my gosh, they're so disrespectful and lazy.

And that is all because of the way the system is set up to operate. Now no one is telling us that because they don't come, we should think of them poorly, but we know we all have these biases, and we're all functioning in this world where when someone doesn't come it's problematic for how we run our services.

And so when I think about the non-emergency medical transport issue, first, it really sets up this dysfunction as it relates to things, you know, inside visits. But it also, like, crams all of these things to these unconscious biases that people have, that lead to like long-term poor consequences as they capitalize and build on each other.

And in addition, we're just not structured in a way that we have policies and strategies to get these families what they need. So if I'm working a shift job, but I have to take an additional two hours off, just because I can't trust that they're going to come when I need them to come, that's two hours of income that I don't have.

And if I am a person living in or near poverty,

that two-hour income could be the difference between paying my electricity bill on time, right? And so this over and over capitalizes and grows, so that families are really shoved into this rock and a hard place situation where their choices just are not tenable.

And we, as providers, start to think well, you know, they're so irresponsible when it's not that at all. So when I think about attendance policies it's often something like, you know, three strikes you're out. You don't show, you come late, you don't get the care, okay.

Or if you come a half hour late, oh too bad, you showed up, but we're not going to see you. We all know that it's set -- these attendance policies are not set up to be punishing, but they end up being punishing. And you know as overhead is expensive, and clinics want a lot of through-put, one of their solutions is a three strikes you're out policy, but who does that impact?

When care is foregone, and transportation in this example is a problem, I can tell you exactly who it impacts. It impacts the kind of kids who showed up as a positive newborn screen. Kids who are more medically complicated. It shows up as impacting the kids from minoritized backgrounds. It shows up in families who have limited financial resources.

So why is it? So it's the poor families who report it the most, and we have to really understand. It's like this why is a complex why. And the conclusions that we jump to are not always the right conclusions. And the way that impacts children and their families long-term could really be detrimental.

So when we put on our health equity glasses, we really need to recognize that there's a lot of injustice baked into the system. There are biases that we all hold onto, and that lead someone like Martin Luther King, Jr. to say of all the forms of inequality, injustice in healthcare is the more shocking and inhumane.

And so for us, it's a call to action. To say we've got to change the way we do things, and the kids get the follow-up they need, they get the services they need. Those services are easy to access, are family centered. Meet their needs. Give them what they deserve, so they can be as healthy as possible and achieve health equity.

All right guys, I think that my time is up, so I'm going to stop the share of my screen, so we can move on to the next portion.

Committee Discussion

DR. CALONGE: Thanks so much for your time, Dr.

Houtrow, that was just super. And describing it the way you did was exquisitely done. I do a lot of health equity work, that's what I was doing before I came back over to the Health Department, and the School of Public Health.

And I think applying the same issues that we see at all ages in terms of challenges to equity apply specifically in the area of newborn screenings is very helpful to this Committee in thinking about moving forward, so thanks so much for your time. We're going to start out with discussion, and as usual I will call on Committee members first, for questions or comments, and then we'll turn to organization representatives. And first up I have Jennifer Kwon.

DR. KWON: Hi. I'm Jennifer Kwon, Committee member. That was a wonderful talk. As somebody who directs a neuromuscular clinic, I feel like I live a lot of the issues you described, and so of course I am impatiently thinking about solutions.

I do really appreciate you bringing these things up. I also want people to know that I think the greatest source of health equity is our medical system. You know, the medical system that we live and work in is really the biggest barrier that I fight in terms of trying to get people care.

So I'm interested in your solutions, and I also would like to highlight, I think, one or two points, key points for me, which is that newborn screening can improve diagnostic health equity in terms of allowing children who have serious disorders to be identified in a timely fashion.

But it may not result in the improved outcomes that people hope for without real improvements in our existing structure of care. But anyway, thank you.

DR. HOUTROW: I really love that, and I think that your point is so well taken about like we have done so much to improve equity in diagnosis because it's universal, right, so we're capturing almost everyone. But universal things don't always work as well for all groups, right, unless it truly is wholly universal. It's really you will have these inequities.

And I think we should celebrate the achievements, like universal screening, but then recognize that where we have to move the needle in the actions we take are different for different groups. I mean, I didn't mention rurality, but rurality, coming from a rural locale, to try to come to a multi-disciplinary neuromuscular clinic, that's a huge undertaking, right?

And I think our kind of attitudes around what

we expect families to be able to overcome are just unfair to families. Our health system is not structured in a way that is easy to use and navigate. You know, I have chronic health conditions. I've had them my whole life. I have been a user of a high-level amount of healthcare my entire life.

I'm a doctor. I have a Ph.D. in medical sociology, and an MPH in health policy and management, and I struggle to assure that I have adequate access to the care that I need, right? Just imagine what it is like for families who don't have the capacity or the ability to navigate the way that I do because I work in a health system. And so I think we need to be thinking about what it means for us not to be like simple work that we do for ourselves, which is hard work, but needed work about how we perceive when families don't show up, or follow-up is lost.

And then also what are the mechanisms by which families will tell us what would help them get to the appointments, right? Like we need to build the equity activities with the people who will benefit and need those equity activities. So like me just sitting here typing on my computer and Googling things is not the same thing as engaging ways with people with disabilities and chronic health disease in the planning

of the activities that they need, so that they can get the care they need.

There's so much inequity in healthcare, so many opportunities, but like we need to really for my big solution point is that and the disability rights movement talks about this, nothing without us. Like why don't we go to the people we need to help to figure out how they need to be helped?

DR. KWON: So what I'm hearing you saying is engage with my population. So actually, and I think that's wonderful to really talk with people who are living it, and then take their words to people who can make the changes. Thank you so much.

DR. HOUTROW: Thank you so much.

DR. CALONGE: I really appreciate you mentioning rurality. You know, from Colorado, that's a daunting task for many of the families whose children are identified through newborn screening, and I know Shawn could talk to that because he lives it, as do others at the Children's Hospital, so thanks for looking that up.

We have about 20 percent of our population that lives in rural or frontier areas for access to care, especially specialty care is just not available, it's challenging. So thanks for mentioning that. Let me

turn now to Cindy Hinton.

DR. HINTON: Hi. Cindy Hinton. Thank you so much for that as a fellow medical sociologist. I really appreciated that. And the one thing that it really brings up for me, and that I think is so baked, also baked into our system is the expectation that is put on people that they need to make individual solutions to things that are environmental problems.

And I mean you see I work in public health. It's like you see that in public health. We are really, as a society, geared towards how do we help you, family, you know, person make these changes to improve your health when so much of it is totally out of an individual's control? And when you compound that with the inequities that, you know, are laid out in terms of complex care, it really is just stunning.

I mean, no solutions offered here. I just really appreciate your presentation, and you know, encourage us all to really think further upstream when we're looking for solutions.

DR. HOUTROW: Yeah. I think that that hits the nail on the head. By the way, I love your wallpaper. I think that this individual solution thing, right, that's really how healthcare operates, but it should not be how we think about optimizing health for a population,

right. So all of the people that live here, and in a really kind of rugged individualism attitudes in the United States that are deeply embedded in how we think about ourselves as a country and a people, ultimately don't end up serving communities so well, and pit one individual's needs against another. Kind of a very detrimental way.

And the thing about health equity is that when our society collectively achieves more health equity, that's better for all of the things that we might want to do as a nation. And so it's not about like oh, I'm not going to get so much of a piece of a pie, or whatever service it is. It's about really understanding that when we all are healthier, right, especially people who are most at risk for poor health, that we, as a society, function better.

And that it's a value to have health in our society, not just a value for certain subsets of people. And I appreciate that there aren't easy solutions, but I think as a professional, you know, I'm doing the day-to-day work with my patients to try to help them be as healthy as possible in my clinic as a doctor, but I'm also thinking what are the things that we need to be changing.

And your term "upstream" really, you know,

feels insurmountable, but things are only insurmountable if we deem them to be insurmountable, and I think we have real opportunity collectively as organizations as people in the field on the ground to kind of work together for collective action.

DR. CALONGE: Christine?

DR. DORLEY: Thank you, Dr. Houtrow. And I must say that I love your glasses being a fellow glasses wearer, I'm always looking for new, exciting frames. But anyway, I wanted to ask, I know many newborn screening programs are incorporating additional testing, like molecular services to try to improve health equity in time to diagnosis.

And we are in the age now where long-term follow-up is becoming more and more developing and creating those types of programs. So I just wondered, you know, what thoughts do you have for programs that are developing long-term follow-up that we can promote health equity in those programs for those infants that are identified by newborn screening?

DR. HOUTROW: You know, I think one of the things that often happens is we're like oh, well, we should monitor the social determinants of health, and collect that information. That's great, totally smart to do if we are willing to intervene on it, right?

So if that changes that we don't just give everyone the same thing. So collecting the social determinants of health information can help tell you where your choices for intervention may need to be different, may need to be amplified, may need to be repeated. So you know what? It would be so much easier for my families that use non-emergency medical transport, than if that service was actually robustly funded and got the families what they needed in a way that meant that they could keep the appointments, and that they didn't have wage loss, right?

There's things like FMLA, for example, that when you have three part-time jobs that are shift jobs, you don't get any FMLA. So recognizing the realities of families' lives will help tailor the kind of interventions that you need to do to get them what they need.

So maybe what they need is not to come into the office, but for someone to go to them. Maybe what they need is co-located services in a way they haven't been co-located before. Maybe what they need is like once they have been recognized as income eligible for one program, they automatically get enrolled in all of the things for which they would be income eligible for. Right?

There are things that we can be thinking about, and then in a world where that automatic enrollment doesn't exist, right? Even in the healthcare space, we know things like housing impacts health, and obviously for kids, nutrition impacts health, but how much time do we spend assuring these families are getting actual nutrition?

I sometimes write prescriptions for food for kids because I can medically justify it. But I shouldn't have to do that, right? We should have structures in place. And when we're thinking about families that are kind of are marginalized and are vulnerable to the effects of poverty and discrimination, those are the places where I think we tailor when we discover them, and then we work extra hard at trying to kind of undo the systematic way these things are operating.

DR. CALONGE: Jeff?

DR. BROSCO: Equity is one of the four pillars of the MCHB's strategic plan, and it's one of the four cornerstones of our blueprint. In fact, Dr. Houtrow was the first author on our blueprint equity paper. And I just wanted to remind folks. We talked a little bit about this at the May meeting, and yesterday Ned mentioned our Propel and Excel grants, and we also have

announcing the Co-Propel ones that will be coming out soon.

What we talk about in that is in the newborn screening how we think about equity is that we want to, as Christine just mentioned, start looking at longer-term outcomes. And even if that's just to age 3, and whether children are thriving, and families are doing well.

And we're asking that states begin to start thinking about how to report that data, and whether it's in EDHI or in other worlds, not just as the across the board how everyone in the state is doing, but also then disaggregating the data based on whatever the historically underserved roots are in that state.

So whether it's rural residents, race, ethnicity, limited English proficiency, a variety of things. And then the states can say what's our strategic plan for making sure that all groups are getting to the highest level? And states aren't asked to be doing this alone, we also have a whole range of grants that are blueprint-specific.

I'm not going to go through all of them now. We recognize that the issues that might be for a child with cystic fibrosis or sickle cell are probably not that different as they are for a child with spina

bifida, or cancer. So trying to work at a state level to improve systems of care is another key part of this. So I just want to let you know that we are thinking about this, working on this, and so grateful to have Dr. Houtrow present today.

DR. CALONGE: Thanks, Jeff. Bob?

DR. OSTRANDER: Thank you. Robert Ostrander, AAFP. I have three brief comments, and two of them have been covered, so I would like, first of all, though, to talk about the fact that you pointed out that newborn screening can improve equity, but I wonder if equity should be one of the things that we consider when we're looking at nominations?

For example, as you were talking, I did a quick literature surfing for the article from March of last year that showed the difference in time to diagnosis from symptom onset to diagnosis and treatment by race and ethnicity, and it was significant for Duchenne muscular dystrophy.

So should it be considered Duchenne muscular dystrophy, adding it to the RUSP, should we consider the fact that using the word screening would reduce that equity? So that was one question to the point. The other comment, and it's been brought up some, but I'd be interested to hear you, Dr. Houtrow, speak somewhat

boldly and specifically about systemic solutions.

You know, there are a lot of, you know, DEI sort of talks at most meetings that I'm part of now, and they all seem to be focused largely on convincing us that there's a problem. And I think we all kind of know that. And I, you know, appreciate the data, and the talk, but, you know, what are the systemic solutions?

And be bold with your thoughts about that, I guess, would be my second comment. And my last comment is rural health is a big part of my world, and what I do, although it says New York, I'm from western New York in the middle of farm country, and lots of Mennonites folks.

And the rurality issue is a big one. There always seems to be a footnote to these things, or not brought up at all. You know, I don't see the data, and when I bring it up at meetings, honestly I sometimes get pushback because, at least from our area, and rurality of this, they're white people, and any time you start to bring up the possibility that there may be this sort of thing going on with white people, sometimes you can get pushback, and you know, feel like we're culturally appropriating or something.

So I would love to see more data on the rurality issue, and also, you know, just people to be

aware that, you know, pushback on that is not necessarily appropriate.

DR. HOUTROW: Yeah, I think that's a super important point. And we often see maps that divide things by states, right? But that doesn't still get at the reality thing that we see. And I think there are definitely ways to look at data from, you know, how far away you live, geocoded with more finesse than, you know, what data the data that I showed today. And I think is really important to acknowledge how much of a barrier distance to care can really mean.

And your point about delayed diagnosis by marginalized racial groups, man, it's really profound. It comes up in kind of every kind of diagnosis that there is. I mean, you know, things like autism delayed diagnosis by years, and that delay in early childhood could really be detrimental for improving outcomes for those kids, and the stress that that creates for families.

And you're so right, as we often talk about like here's a problem, here's a problem, can I show you the problem, but don't talk so much about solutions. So you said people who are not part of health equity blueprint that Jeff was just talking about. And we made points in that about, you know, we could reduce child

poverty by 50 percent, sustain a reduction in child poverty, and have that be financially beneficial.

So the NAM, National Academy of Medicine, which was the Institute of Medicine before a few years ago, came out with a report about reducing child poverty. And, you know, the data is robust that we can reduce child poverty in half in a decade, and end up with more money than if we didn't do that.

So that, so child tax credits, for example, which were a part of what happened during the pandemic, food assistance services, and a whole host of things. And what that report actually did was come up with like if you do this, plus this, plus this you'll get this. If you do this plus this plus that, you'll get this other thing.

So it's really great, and I can put the link to it in the chat in a minute, great report. And then the other thing is that we really, you know, we have not, as a nation, signed on to a bunch of things that United Nations set forth. So, for the Rights of the Child, for example, we have not signed onto that.

We have also not signed on to the Rights for People with Disabilities. And so those are kind of things that on kind of a political front, we're not engaging with, and I think there really needs to be a

push to go back and really label these problems. And you know, the politics on this, the partisan politics on this are super challenging.

And so I think some about reframing in a way like keeping kids out of poverty isn't about free handouts. It's about taking money for our country, right? So some of that requires some very serious reframing.

But I think that being bold, something like, and Jim Perrin, a well-known pediatrician out of Boston, former American Academy of Pediatrics President, recently wrote, like, why have we not federalized Medicaid? Why don't we make sure all kids get EPSDT services in a way that doesn't -- isn't chock full of prior authorizations and denials and whatnot because it will be cheaper to operate it on a federal level than it is to do it the way we do it now, which is like you see one Medicaid program, you see one Medicaid program, right?

And that's not good for the people that it serves, and it's very, from an administrative perspective, adds complications that we don't need. I often think about, like, what is the thing we give? Oh, let's put in another care coordinator, or another navigator. Why don't we just make the thing simpler?

Like, not make it more complicated, make it simpler.

And so, I think there's a lot to be done, but we have to get rid of whatever perverse incentives drive things to be the way they are. And I wish I had a magic wand to do that, but I do think we need to be bold, you know, acknowledging the depth of the problems, and where the solutions lie at fundamental levels of how things are organized.

DR. CALONGE: It's interesting that the long-lasting legacy of the Grand Compromise still shows up today, especially in terms of how we provide healthcare for our children. Natasha?

MS. BONHOMME: Great. Thank you, Natasha Bonhomme, Genetic Alliance. Again, as everyone has said, a wonderful presentation. I have three things I want to comment on or note. I really appreciate that you spoke about family partnership, and how important it is to hear from families, and to really understand where the touch points that would make an impact in their lives, and really getting into care.

We, through our national genetics education and family support center, recently brought together family leaders from across the country, who are very active. And when we surveyed them, they -- only 22 percent said that there were enough opportunities to serve as family

leaders in the genetics healthcare delivery system.

So I think that shows that even our most engaged groups have struggles to really figure out I have this passion, I have this energy, I have this opportunity; where do I put those efforts? So I think that just kind of makes it like what you were saying around partnerships.

I also wanted to note that though we talked a good bit about rural, me being here in D.C. in an urban environment know that there are people who it takes over two hours to get to maternity care that's four miles away because of how transportation is.

There are a lot of efforts going in, but I think it's important to remember that yes, there are definitely struggles in rural areas, as there are in our urban settings and environments. And then lastly, and maybe this is something you could comment on if you would like, you know, we often use the word universal to talk about newborn screening. And yet it isn't, completely, when we think about how every state has a different panel, and how and what conditions are screened for are different.

And it was, I don't know if funny is the right word, but when you were talking about Medicaid, I was like, oh, that's kind of how we talk about newborn

screening. If you see one newborn screening program, you see one newborn screening program. And you know, the work that Amy Gaviglio and Erin Goldenberg and Beth Tarini have done, and I think have presented to this Committee, have talked about kind of the differences that we see.

And we don't have to wait to see a child at three years old. Like the differences are from the beginning, or even before the beginning, and how there are differences in state newborn screening programs, and then differences in the type of data we can get, and then it just kind of layers on from there.

So I just wanted to kind of bring that piece too, if we're really talking about equity and newborn screening, it's not just starting at the point where the family comes in. It's really from even before then, so. But again, thank you so much for your work in this presentation, and I hope we can get updates and refreshers on it because it's really critical.

DR. HOUTROW: Well, thank you so much. And I love your point about, you know, you've seen one newborn screening program, you've seen one. And you know, there are decisions that were made long, probably before many of us came to sit at these tables about which ones were included, and then how do we act after one is positive?

And those differ by states also.

And those are kind of where our priorities are, are deeply entrenched in our kind of cultural value and lens, right, that drive decisions about what we test for. Oh, this one is costly, but you know, very amenable to early intervention. Okay. So, yes, we want to, what are the things that we use in our decision making about what goes in and what falls out are all really culturally and politically mediated.

And I think we all have to recognize that those things like, well, you know, we did this evidence-based thing, but that evidence-based thing is built on, you know, lifetimes of inequities being baked in to how we think and operate, and who has the power to decide things, right?

So when you look around rooms of places where decision makers are, they often don't share my gender, and they also certainly don't share my disability status, and you know, I have incredible privilege because of the color of my skin, and my nativity that I have English as my first language, and I come from a family of people who were educated.

But we often do not locate our privilege when we assess evidence and data, and where that evidence and data came from. And so I really love the kind of what

work needs to be done to do some equity alignment, even about what's included in newborn screening.

And then making sure that people who come from typically unheard and oppressed groups are engaged in the evaluation of that data, and that we are taking a critical eye of what bias was already baked in, right, and what structures of how things were done before leads us to conclude certain things.

And it's hard, incredibly hard work, but I think it's necessary work for us to get to the place where we have our, you know, full equity lenses on. And I really appreciate that point because it talks about universal and not being universal at all. And how when you call it that it isn't really that.

And I think naming things, calling it out, I'm so glad you did that because we have to do that. In a term like universal sounds universal, even when it's not, so I thank you so much for that.

DR. CALONGE: Susan Berry?

DR. BERRY: Thank you so much for that presentation, which I found to be really enlightening about the honesty we have to have as we approach many of our areas.

The comment I wanted to make was I was thinking about our discussion yesterday which was about Duchenne

muscular dystrophy. And we were talking about the definition of Duchenne and saying, and we should have a genetic definition. It should have a genetic test that matches with it. And I thought to myself, well, who's going to pay for the test? How's that test going to be done? It's expensive. How do we make sure that's truly available to every child, so that we have an equitable and appropriate definition as well?

I mean, just from the core beginning of what is the disorder. And that's also true as we think about adding disorders and say we want to add this disorder because there's a treatment. It costs $1 million. How do we make sure that we think about that impact, and make it as fair and as achievable as possible for all the babies?

And so, you talked about it being baked in. It kind of is baked in. Our science is way ahead of our ability to provide equitable care, and we don't consider it as actively as we probably deserve to, as our patients deserve when we are considering those options and care. So that's my comment. It's not really a question. It's really an editorial.

DR. HOUTROW: No. I really appreciate it because you know, I take care of kids with muscular dystrophies in my practice. And you know, kind of the

exciting things are these really new medications that are available that are just so lifechanging. But you know, one of the things that, as someone who has studied in public health, and like, you know, it doesn't cost that much money to make sure people have some of the basic things that they need.

And in the long-term consequences of poorly managed, under-diagnosed, under-treated conditions that relate, the reason those things happen is how we have biases baked into our systems, and how we can't get families just the basic things they need to thrive in their communities, also makes me think about like, you know, which of the big talks about where and how we use our resources.

Your other point is like, well, we need to have actual diagnosis. When I started training, we were doing EMGs on kids with static conditions to diagnose them. And now, thank goodness, we're not doing that. But we also have a serious problem in which, you know, things like delayed speech is thought of very differently from a white, upper middle class family's child than if it occurs in a black family's child who are more likely to be poor.

And that we're just like oh, you know, they'll catch up. It's a completely different discussion, and

our kind of fundamental biases are baked in about those kind of things play out in very negative ways. And so, you know, I really love the kind of thinking that we're doing here, and this group keeps -- I mean, this is one of my favorite things, is to have these kind of discussions and hear all of your guys' wisdom.

And you know, our ability to, like, hone in on one specific thing, and then spread out the lens to see all of the bigger things. And our responsibilities to the people that we serve to do that, to go deep on let's say muscular dystrophy, like you just did. And you know, pull out to see the bigger milieu.

I think is the strategy that we need to use organizationally when we're having these kinds of discussions. Even when it is a targeted discussion, we need to then blow out our lens and think much bigger.

And so I really love the opportunity that you just --for me to be able to say that in response to something that's so near and dear to my heart in the place of muscular dystrophy, and helping those families go through a situation that that diagnosis can be so incredibly devastating, but have you know, kids who have such full and rich lives, and that we help be as capable as possible.

So thank you. I really appreciate that.

DR. CALONGE: Michele?

DR. CAGGANA: Hi, everyone. Thanks for that talk. It really brought home a lot of the things that we think about from the newborn screening perspective. And as part of newborn screening, the short-term follow-up piece, you know, is sort of when that diagnosis is made and is that baby in care. And as Chris mentioned, historically that's where it cuts off, and then we go in to long-term follow-up.

So I'm wondering if, as a group, newborn screening programs could think about how to build an equity piece into that and really track those factors down. APHL has some data from their NewSTEPs system that talks, that looks at kind of diagnosis broken out, and there was a presentation to this Committee, I don't know when it was, showing that there are delays in diagnoses even though programs get those results out right away.

And I think it's really hard from our perspective to kind of look out and see or make changes to the way we do things in order to be able to improve that. But I think one of the most powerful things that you mentioned to me at least, was this sort of the value of -- the value added, right?

So, like you mentioned, you have a family, they

missed their appointment, they missed work. They can't buy something. They can't pay a bill, there's all this trickle down. And I'm wondering if there's any way that I think the greater public can appreciate that more. Is there any way that we would be able to, you know, make that more granular, I guess, and sort of show that we all suffer when people don't show up, or you know, when parents have trouble getting to work, or getting their child to their appointment.

And I think really thinking about collecting better data from a long-term follow-up perspective can really help us with framing this to departments and get people thinking more about this, rather than just throwing money at things, and not really, like you said, stick it where it needs to be.

DR. HOUTROW: Yeah. Ultimately, that's a money-saving intervention.

DR. CAGGANA: Right.

DR. HOUTROW: Is to not just throw money at the problem and see, you know, what comes out of it. And I think, you know, I think there's like this linkage to what happens long term. Obviously, the data is very important. And then, how do we create the right bridge? And not everyone is going to name, you know, the bridging from, you know, the diagnosis and delivering of

that information to the long-term in the same way.

And so how do we tailor that in a way that meets people's needs, and is not burdensome for people? And, you know, I think also the messaging part is really essential as we think about the granular aspects, as an example that I've given, and you brought up about attendance policies. Well, there's a whole bunch of incentives as to why attendance policies exist that way, and they have really negative consequences for families. And they also have negative consequences for, you know, the system itself, and that it needs revision.

But the other thing is that people really do because all of us have experienced barriers to something like that are you know, a car tire. You got a flat tire when we were driving somewhere, and we missed some sort of like thing that we were supposed to go to, or we're late for our granddaughter's wedding, you know, whatever.

And so, you know, when we contextualize these things as more human experiences, versus like, oh, well, I see that as a problem of this group, right? Because these are real. We all have universal -- huh, the word universal again, experiences where things don't go the way that we always expect them to despite our best intentions.

And I think also kind of believing in our best intentions even in the face of an inequitable and structured system that doesn't allow those best intentions to be lived, I think is really important as for those of us in the space, holding onto the idea that it's not that, you know, so-and-so's parents are trying to undermine my clinic by not calling me to tell me they're not coming.

There was no mal intent by them. They're not thinking how could I ruin Dr. Houtrow's day today. I think we also really have to hold on to, as people in positions of authority, and people in the service space, is to really hold on to the kindness that comes with understanding people's best intentions, and how hard it is to achieve them.

DR. CAGGANA. Thanks.

DR. CALONGE: So, Karin, we're on time, so I'm going to give you the last brief question or comment. You're on mute.

DR. HOUTROW: You're on mute.

MS. DOWNS: Thanks for that. I lowered my hand, but I didn't unmute. I really appreciated your presentation, and many of the questions I was going to ask were responded to in your response to other people. There are two things that I wanted to add to reflect on,

and I think we're doing a good job of doing this.

One of them is really around developing data systems that look at the system's issues more than, you know, all the data you showed from the National Survey of Children's Health, really is individuals' experience, but not really, you know, what policies are in place that contribute to structural racism for example? What contributes to systemic racism, and what are those measures?

And I think we're starting to work on that, and so you know, I would really -- again, thinking boldly, until we start being more refined at looking at the measuring structural differences and systemic differences, we're going to continue to go back to falling on, well, this person needs to eat better, or that person needs, you know, transportation for this family.

And, again, that's like pulling children out of the river downstream. So that is one thing, and I think I really appreciate how everyone is thinking creatively about systemic and structural measures that are not really based on individual measures, that's one point.

The other point is that, you know, we've sort of danced around this and alluded to it, but I also think that we need to do a much better job of measuring

and looking at implicit bias, and having everyone within the health care system, you know, recognize that we do bring implicit bias, and that may blind us to our privilege, whether it's educational privilege, racial privilege, whatever privilege.

And so, I think, you know, I'm saying two things at the same time. We need to do a lot of internal work, all of us, and that we need to look at the system's piece sort of simultaneously, so...

DR. HOUTROW: I love that. You know, the causative nature of poverty on outcomes related to health and disability is, like, well-established cost and relationship. And yet we don't really tackle it. And I show the associations with our data, there are a number of, you know, people in power around the country who do not believe there's any systemically embedded injustice in anything.

And so I love the point of we need the data that demonstrates where the locus of the problem is, instead of focusing on the downstream differences, and then trying to intervene there. And then also, clearly very important is our own recognition that the world that we view.

In another talk I show -- do you remember a few years ago when the internet was having a problem.

DR. CALONGE: Dr. Houtrow --

DR. HOUTROW: Oh, you need me to wrap up.

DR. CALONGE: I'm sorry to interrupt because we have public comments next, and you can continue to talk for a long time on this topic. I want to just pause and tell you I know I speak not just for the Committee members and the organizational reps, but everyone on the call in thanking you for sharing your expertise, your considerable amount of time and guidance today. It's been great, you've given us a lot to talk about, a lot to think about, and hopefully a lot of things to act on. So thanks for your time today.

DR. HOUTROW: Thank you for inviting me and allowing me to be as long-winded as I was. I really appreciate it, and it was great to learn from all these people in this group who are very dedicated and wise, and I appreciate it.

Public Comment

DR. CALONGE: Thanks so much. I want to move on now. I have eight people signed up for oral public comments, and we will bring you on, ask you to unmute as we move forward. And we're going to start with Kasey Feldt.

MS. FELDT: Hello. And thank you to the Committee for allowing me to speak on my experience with

Krabbe disease. My name is Kasey Feldt, and I am a mom of a beautiful and strong Krabbe angel, Dawson Luke Feldt. Dawson was born on July 16, 2019, and he was absolutely perfect.

As the months went on my husband and I noticed Dawson never gained head control. He often got sick, would throw up his food, lost his laugh and smile, and eventually started having muscle spasms. After months of testing, on February 24, 2020, we heard the words early infantile Krabbe disease for the first time.

Hearing that he would not live past two years old brought me to my knees, and made me scream in pain, devastation and grief. My son passed away eight months later at the age of 15 months old.

My husband and I recently attended the Hunter's Hope Symposium, and what we saw amazed us. We saw children with Krabbe who were transplanted before symptoms, living and happy. A 23-year-old who was transplanted at three years old was performing professional magic tricks that stunned the audience. An 8-year-old who received a transplant for Krabbe's disease just after birth recited the Lord's Prayer. And a 19-year-old who received a transplant as an infant sang One Hell of an Amen by Brantley Gilbert.

I can only wonder if my son Dawson could have

done these amazing things because Virginia never gave him a chance. Newborn screening is crucial for families to have that time with their kids.

I only got 15 months with Dawson, what I wouldn't give to hear him say mama, dada, or I love you. You have the chance today to do something amazing, to give families time. You do not know how precious time is until you are given an hourglass, and you are just waiting for the sand to run out. But with newborn screening and transplant, these kids can live. Thank you.

DR. CALONGE: Thanks so much. I'd like to move on to Phil May.

MR. MAY: Good morning. Thank you for giving me the opportunity to speak with you today. My name is Phil May. I live in Tennessee. This is my son Dylan, who was born in 2004 and died just before his fifth birthday in 2009. Dylan was a beautiful, happy, sweet little baby that was of course loved unconditionally by me and my wife and our two older sons.

After starting to lose some motor skills, Dylan was eventually diagnosed with Krabbe disease at eight months of age. For Dylan that diagnosis came too late for any treatment. The doctors told us to take him home and make him comfortable until he died, and we should

expect that to be between the ages of 13 months and two years old.

We were blessed that Dylan lived to be almost five. During the more than four-year period after his diagnosis, the disease progression was really unbearable to watch. We lost pieces of Dylan slowly and painfully. He lost the ability to move his arms and legs. He lost the ability to eat solid foods. He needed regular Botox injections in his legs to allow us to bend his legs enough to change his clothes.

He regularly vomited his food after we spent hours each day trying to feed him. He was never able to speak. He had extreme recurring seizures. He was often obviously in pain, crying out for long periods of time, and we had no idea how to comfort him. He lost his ability to smile, and most painfully for us, Dylan lost his eyesight.

As painful as it was to watch the disease slowly take our son's life, our other sons, Jackson and Connor, and our daughter Sophie, who we adopted when Dylan was 3, all experienced this same loss. Each of them have complicated mental health issues related to the years of trauma from losing their brother in such a horrific manner.

Had Dylan been screened for Krabbe at birth I

have no doubt he would be living today as a 19-year-old. I often, even 14 years after his death, envision what his life could have been had he been screened at birth and treated. We never saw Dylan on his first date or graduate from high school. Nor will we see him graduate from college, get married or have children of his own.

I'm not basing my confidence that he would be living today on what I've read or heard from others. I've seen with my own eyes many children of Krabbe who were screened at birth and treated. I've held him them in my arms. I've seen them playing with other children. I've seen them laugh, attend school and thrive. Without screening for Krabbe at birth all of these children would have died a slow and painful death.

And because so few states screen for Krabbe at birth, I know many more children who are in this excruciating process of dying at this very moment. I implore you to do all you can to ensure Krabbe is included in the RUSP. No parents and no children should ever have to endure the excruciating loss that we, our other children and so many other families have faced.

You have the authority to save the lives of children with Krabbe, and by doing so you will also be saving parents and siblings from experiencing life-altering trauma, and the lifelong effects of such a

devastating loss. Thank you.

DR. CALONGE: Thank you. Next I have Dietrich Matern.

DR. MATERN: Thank you, Dr. Calonge, for the opportunity to return to the commenting and speak again in support of the nomination of Krabbe disease to the Recommended Uniform Screening Panel. As you know, I have been very disappointed and frustrated at the outcome of last February's meeting.

From the perspectives of the nominators, including myself, which was outlined in the various responses the Secretary and you received since February, all available on the Hunter’s Hope of websites. It has been a surprise how negatively newborn screening for Krabbe was described.

Yes, the history of Krabbe newborn screening did not go as planned, but the Committee did not consider the data in the context of the nominated screening strategy, which uses a biomarker, psychosine, as a deciding factor whether a screening result is positive or negative. Psychosine measurement was not clinically available until 2015, nine years after screening started, and was first employed as a second-tier test for Krabbe screening in Kentucky where it virtually prevented false positive results.

Since then, 9 of the current 11 states screening for Krabbe disease use psychosine. However, most states still struggle to abandon genotyping out of fear of missing a case. I believe that the public health program needs to balance the benefit of a few that need early treatment, and those who are unaffected, but could be negatively impacted by a false positive result.

I think we agree on this point, so how do we improve this? If you consider the history of the ACHDNC, please remember the impact your previous recommendation have made, for example, for better timeliness, screening for Tyrosinemia Type I, and of course SMA.

Without the Committee recommending newborn screening for SMA defined as homozygosity for specific deletion, providers would be inundated with helping stressed families figure out if their newborn is affected with this deadly disorder, or just a healthy carrier. Just as in those instances, you have the power to improve newborn screening for Krabbe disease by adding Krabbe disease to the RUSP defined as reduced health activity and elevated psychosine.

32 percent of U.S. newborns are already being screened for Krabbe disease, but at least one state,

Minnesota, likely adding it within a year. Adding Krabbe disease to the RUSP as recommended would make screening for Krabbe disease equitable across the country, essentially eliminate false positive results, and give affected babies a chance at life.

All this at a minimal cost because most states screening for other RUSP conditions already measure GALC activity because a testing kit includes the necessary agents. In summary, the nominated screening procedure for Krabbe disease is efficient and effective, and states already screening for Pompe disease or MPS can easily add Krabbe disease to their panels.

Relevant follow-up and monetary guidelines have been published in peer-reviewed articles, and the NCC already developed and published ACT Sheets full of algorithms and a knowledge nugget. Experts in the field working with patient advocacy groups stand ready at all times in support also of equity after newborn screening by facilitating timely and appropriately, through the screening of newborns to ensure families receive the most up-to-date information to make the best decisions for their baby and their families.

Therefore, I implore you once again to use your power to finally give all future U.S. children with special needs due to Krabbe disease a chance, no longer

condemning them to the suffering caused by this horrific disease. Thank you again for giving me the opportunity to speak to you, and I'm happy to answer any questions you may have.

DR. CALONGE: Thank you. I'd like to turn now to Joanne Kurtzburg.

DR. KURTZBURG: Thank you, and hi, everyone. My name is Joanne Kurtzburg, and I'm a transplant physician at Duke Health. I've testified in the last two meetings, and I'm back today to share some new information about the nomination of Krabbe disease for addition to the RUSP.

As you know, Krabbe disease was nominated in 2009, and again in 2021. Unfortunately, it was not approved to be added to the RUSP either time. Several criticisms were discussed, and we have attempted to resolve each one of them in a new and modified nomination, which will be discussed later today.

First, ability to diagnose Krabbe disease to newborn screening was challenged, and as you've heard multiple times from Dr. Matern and in the evidence review, the addition of psychosine as a second-tier test for babies screening positive for low GALC now enables definitive diagnosis of babies with the infantile, formerly called early infantile, form of Krabbe disease.

Treatment of these babies with hematopoietic stem cell transplantation in their first month of life was lifesaving, and significantly improves the quality of life of these babies, children and their families, which is a major justification and motivation for adding Krabbe disease to the RUSP.

Another identified challenge was specificity of diagnosis of Krabbe disease through newborn screening. When we nominated Krabbe disease for addition to the RUSP in 2021, we included the diagnoses of babies with the infantile, formerly called early infantile, form of the disease, meaning disease presenting before 12 months of age, and delayed infantile, meaning disease presenting from 1 to 3 years of age as the nominated forms of disease.

Diagnosis of babies with the infantile disease was based on a positive screen and a psychosine greater than or equal to 10 nanomolar, while diagnosis of late infantile disease was based on a positive screen and a psychosine between 2 and less than 10 nanomolar.

The latter category was divided into high-risk and low-risk groups based on additional testing with mutational analysis. We now recognize that the inclusion of the group of babies with intermediate psychosine created confusion and a lack of clarity

around the diagnosis of Krabbe disease. It also had the potential to cause distress for families where definitive diagnosis of Krabbe disease could not be confirmed.

We also recognized that medical management of this intermediate group is not firmly established, creating more uncertainty, both for newborn screening labs, and among healthcare providers. To remedy this situation, and also to enable diagnosis of babies with the most severe and rapid form of Krabbe disease -- sorry -- rapidly progressing form of Krabbe disease where early diagnosis is paramount to enable early treatment.

We've now amended the Krabbe disease nomination for addition to the RUSP to be restricted to infantile Krabbe's disease. Infantile Krabbe disease can be diagnosed through a GALC screening, followed by second-tier testing with succus. Babies with a positive screen and a psychosine greater than or equal to 10 always have infantile Krabbe disease.

The parents of these babies should be notified, rapidly counseled, and given the opportunity for urgent referral to a transplant center for workup for future transplant, or gene therapy. Additional questions were raised about whether babies with infantile Krabbe

disease referred for transplant during their first month of life really have infantile Krabbe disease.

I can assure you that all of the babies with high psychosine have active signs of active disease in their first few weeks of life. Active disease includes white matter changes on MRI, abnormal nerve conduction studies and elevated protein and psychosine in the spinal fluid, accompanied in some babies by changes in EEG and auditory and vocal responses.

This information was published in 2005 and again in 2022 in two serious reporting outcomes of transplant in babies with infantile Krabbe disease. As you've heard many times, babies with infantile Krabbe disease rarely gain milestones past smiling. They never develop head control or sit, crawl, stand, speak or walk.

Within a few months of age, they have feeding problems, extreme irritability, seizures, blindness and spasticity. Babies who are transplanted in the first months or so of life never develop irritability, are cognitively normal, and gain developmental milestones, albeit at a slower rate than typically developing children.

I personally have treated and followed over 30 of these babies, some of whom are now teenagers and

living into their 20's. While I do not deny that some of these children have motor disabilities affecting their ability to walk, these children are happy, social, attending school. They have friends, they use computers. They can see, hear, read, communicate, and have a much better quality of life than untreated babies who suffer extensively, only to die in a median of two years of age.

As you will hear today, we submitted the revised nomination of Krabbe disease for reconsideration on the expedited pathway for addition to the RUSP. The revised nomination restricts the diagnosis of Krabbe disease to newborn screening to the infantile form. Diagnosis of this form of disease can be accomplished rapidly with the GALC screen, and second-tier psychosine testing on screen positive babies.

If the psychosine is greater or equal to 10, the baby definitely has infantile Krabbe disease. Parents can be informed and given the option to be referred for further evaluation and treatment. This strategy will greatly reduce the suffering of these babies and their families who will otherwise experience it as you have heard from many of the speakers today.

Additional benefits include elimination of confusing results and unclear pathways for a follow-up

for risk of later onset of Krabbe disease, eliminates diagnostic odysseys for parents of affected babies, and provides access to genetic counseling.

In summary, I strongly encourage the Committee to vote today to allow reconsideration of the revised nomination for infantile Krabbe disease on the expedited pathway agreed to during this meeting. I also sincerely hope this reconsideration will result in a positive vote to add infantile Krabbe disease to the RUSP in the near future. Thank you for your time today.

DR. CALONGE: Thank you. Now I would like to call on Elisa Seeger.

MS. SEEGER: Dear Chairman Calonge, and members of the Advisory Committee for Heritable Disorders in Newborns and Children. Thank you for the opportunity to provide public comment. My name is Elisa Seeger, and I am the founder of the ALD Alliance. Newborn screening advocacy and education are at the core of our mission, along with providing support to families affected by ALD, a disease that is not equally screened for in our country due to the disparities that exist in our country today.

I would like to draw attention to the advocacy work that our coalition has been doing to end death by zip code. As many of you here know, the state where a

baby is born determines which conditions they are screened for, leading to inequalities across the country. To end death by zip code, the country must prioritize complete RUSP implementation in all 50 states.

We have consistently heard from state labs and the countless research done that funding is one of the major barriers to efficiently implementing newborn screening conditions. The CDC, under their newborn screening quality assurance program, and HRSA, both offered funding opportunities last year through grants intended to help states to build capacity to support implementation of the RUSP conditions.

Today, as this community looks to the future with Duchenne and Krabbe in mind, I implore this Committee to allot more of its time and resources to finding ways to fund our country's newborn screening labs. As a foundation that has lobbied tirelessly for the CDC's enhancing disease detection in newborns, building capacity in public health laboratories grant, I applaud HRSA for the recent newborn screening Propel grant opportunity.

However, there is a need to address this to take into account the birth rate and individual states to correlate with the funding that is available. One

state looking to add MPS II, a condition recently added to the RUSP, estimated a cost of $750,000 to complete implementation.

This state received approximately $345,000, about the same amount that all states were awarded. $120,000 of the 345 must be used for the state's follow-up program. As per the grant, it cannot be used for implementation, leaving the state left with $225,000, which will only cover 30 percent of their costs to add the new RUSP approved condition.

We are asking our state labs to do the impossible. What purpose does the RUSP serve if states are not properly funded to add the recommended conditions? Many are so short-staffed that they cannot even designate an employee to apply for the HRSA and CDC grants.

For this reason, I am presenting three asks today. Transparency. The CDC has a website dedicated to their grant program that shows the history of the grant, awarded states, amount awarded, and what the funding will be used for by the state. This allows organizations like ours to help target our advocacy and work with state follow-up programs to provide education to their newborn screening follow-up programs. We ask that the same be done for the HRSA Propel grant.

Number two, smarter funding. It is apparent that this new grant opportunity was put together with very little involvement from our country's newborn screening labs. Please include them in the process. $345,000 may be appropriated for some states, but the needs of our labs vary greatly across the country, as does the cost of implementing a new condition.

Accessibility. As mentioned previously, many states do not have the resources to devote an employee to grant writing. It may be a skillset that is missing altogether. A one-hour virtual meeting is not enough to prepare a lab to write a grant with personnel that has no experience writing grants. Grant writing workshops should not only be provided, but incentivized.

It is already a difficult notion that labs should have to apply for funding to do their jobs. Let's make it as easy as possible for them to do so. We will continue to push for more federal funding for states and their newborn screening programs, and hope that state lab engagement and newborn screening process continues, as their voice and hard work is vital for ensuring that geography does not dictate life and death for newborns.

I would be happy to help meet at a later date regarding these requests, and share our thought processes as an organization that has dedicated much of

our time building relationships with our state's newborn screening programs, the dedicated individuals that make newborn screening a reality. Thank you for your time today.

DR. CALONGE: Thank you, Elisa. Next I'd like to call on Dean Suhr.

MR. SUHR: Yes, good morning. Thank you, Chairman and Committee members and other attendees for this opportunity to speak. I appreciate your continued presence on our journey as we continue to work to bring early diagnostics and therapies to MLD families across the United States and around the globe.

At our recent MLD family conference last month, we hosted our traditional butterfly memorial ceremony. We recognized and honored over 60 MLD loved ones lost since our last gathering, many of them would still be with us if we had diagnosed them pre-symptomatically.

I'd like to give you a couple of quick MLD newborn screening updates. MLD Foundation started working with gene therapy researchers in Italy in 2005. We started working with the Advisory Committee to learn and to positively impact the MLD newborn screening ecosystem about 2011. About that same year we started working with Professor Gelb on an MLD newborn screening assay.

I'm pleased to share that MLD gene therapy was approved by the EMA in December 2021, almost two years ago, again the therapy was developed there. That's why the first approval was there. The outcomes for this gene therapy have been extraordinary, and many are saying curative.

But as a rare disease, pre-symptomatic diagnosis is generally not possible without newborn screening, so most families don't have the opportunity to access that therapy. The FDA BLA, the biologics application submission was completed over the past few weeks by Orchard Therapeutics here in the U.S. It has a rare pediatric disease and regenerative medicine advanced therapy designation from the FDA.

The sponsor organizations requested prior review, which if granted would put the therapy on track for a potential U.S. approval the first half of 2024, next year. A publication recently came out last month or six weeks, with algorithm to determine the form of MLD using the babies' variant data, and the genotype phenotype scaling matrix. The authors are reporting a 76 percent accuracy across nearly 500 patients, and the matrix identifies how to deal with the VOUS, the variants of unknown significance, as well.

This is really important because, as we have

discussed and heard, these meetings and plenty of other meetings, we need to know how to properly refer patients depending upon the form of disease that they have. We're adding advocacy and family experience to the scientific basis of this publication to increase the accuracy and expand some of those VOUS and some of those conclusions.

76 percent accuracy is awesome. We'd love to see that in the 80s or maybe up to low 90s. The MLD RUSP nomination is very close to a formal submission to this Committee. And adhering to your requirements to have an approved U.S. FDA approved U.S. therapy, the nomination will be submitting roughly in sync with that anticipated approval.

In addition, four babies have been identified to date through current newborn screening pilots that have screened well over 100,000 babies, so we hit those checkoffs. It's a good therapy for babies, and soon an approved therapy.

RUSP alignment: I want to shift gears and acknowledge the progress of the MLD Foundation, and the many organizations working with them and the families that are participating in the process to advance RUSP alignment. RUSP alignment sets a timeframe and/or a process for a state to consider the review, in some

cases, actually to implement the screening of a new RUSP addition on a specific timeline.

I was the catalyst for the language in California's 2016 Senate Bill 1095 that started their efforts, and I'm just thrilled with the progress they've made since then. In the last month or so they passed Texas RUSP alignment, bringing their RUSP alignment state count to 11, covering 47.5 percent of U.S. annual births, almost half of the country is covered by RUSP alignment at this point.

In parallel, I'd like to give congratulations to the Illinois-based Evanosky Foundation, which brought a 10-plus-year lysosomal disease effort to conclusion with the passing of Illinois Senate Bill 67, which gives MLD RUSP alignment-like status, so this is a singular disease legislation. With this legislation MLD newborn screening, once approved, will cover more than 50 percent of the babies, about 51 percent of the babies will be expedited to implementation.

I'd like to make a quick comment about the review process of the Committee, again, just to encourage additional consideration and thinking. We've talked a little bit about this yesterday. The newborn screening process is purposely optimized and focused on quality of life. Quality of course, but also life. But I

want to ask that you consider, what about death? Not just death, and the negatives potentially from therapy, but death when there is no diagnosis or no therapy.

Not all disorders are fatal, or fatal quickly. MLD happens to be the infantile, late infantile form happens to be that. But death when you can't get diagnosis is death, and that's just a significant thing that you all need to consider when you do not review a therapy, or if you're looking at the positives, you need to add that to the negatives.

So death should be part of that net benefit. Dr. Bailey, former member of this Committee, published a new viewpoint article this week with a core discussion focusing on net benefit. I'd encourage you to take a look at that document. And finally, the Committee approved yesterday the review of DMD.

We'll be talking about a re-review, an expedited review of Krabbe. There's reviews in process, and who knows what else will be submitted between now and when MLD comes on the docket. I remain very concerned about the expert advisory group's capability and capacity to handle multiple simultaneous reviews.

They've shared that they can handle two, but can they handle three, or can they handle four? And we need to keep thinking about that. There is a tsunami of

gene therapies coming along. You will see a dramatic increase in nominations, and I'd like to help you, and encourage you to consider your capacity.

Thank you for the time to speak this morning. I appreciate your hard work.

DR. CALONGE: Thanks Dean. And now I'd like to welcome Kimberly Widner.

MS. WIDNER: Good morning. Thank you for time. I'm Kim Widner, and I currently live in Omaha, Nebraska. One of the worst days of my mothering experience to date was the evening after my daughter Bailey received her diagnosis of congenital cytomegalovirus, or cCMV.

That evening when I was tucking her into bed, I sat with my four-year-old daughter and explained to her that if she were to wake up in the morning and wouldn't be able to hear mommy, it would be all okay. We would get through it together. Bailey has single sided deafness due to congenital CMV, along with bowels loss, ADHD, brain calcifications, which we are still currently anxiously awaiting to see how this will impact her learning as she matures.

Bailey was not diagnosed with CMV until she was age 4, when she was able to tell me herself that she had a loud ear and a quiet ear. Her hearing was progressive when she received her diagnosis. We are still currently

monitoring her other ear to see what her hearing levels will be.

She now has one Cochlear implant to assist hearing in her deaf ear. Congenital CMV affects over 30,000 babies every year in the United States, making it the most common congenital viral infection. One in 200 children are born with cCMV each year, which equates to one child being permanently disabled from CMV every hour.

Common disabilities caused by CMV include hearing loss, vision loss, mental disability, brain calcifications, cerebral palsy, seizures, developmental and motor delays and even death. More children are disabled by CMV annually than any of the other following congenital conditions: HIV, Down Syndrome, Sudden Infant Death Syndrome, or SIDS, spina bifida, fetal alcohol syndrome, group E strep, and Rubella.

Our family story is so similar to every other CMV's family that I have met through our journey. Why did we not know about CMV? Why didn't any doctor discuss this virus with us?

I was worried about the impacts of kitty litter, deli meat, but never once was CMV, the prevention methods, and/or screening and testing ever discussed with us. I'm here today asking you, and

pleading for more to be done so future moms and dads and children won't have to unnecessarily battle the impacts of CMV for their lifetime.

I ask you, if you were a new parent whose newborn was just diagnosed with hearing loss, only to find out that your child was exposed to a virus in utero, that no one in the medical community warned me about, what would you do? Thank you for your time today.

DR. CALONGE: Thank you. Last, we have Dylan Simon.

MR. SIMON: Good morning. My name is Dylan Simon, and I serve as the Director of Policy for the EveryLife Foundation for Rare Diseases. On behalf of the EveryLife Foundation, I would like to thank the Committee for providing me with the opportunity to provide updates on our relevant newborn screening initiative program.

EveryLife's newborn screening initiative is focused on ensuring babies receive lifesaving treatment opportunities through early diagnosis with newborn screening. I would like to thank the Committee for providing me with the opportunity to provide updates on our relevant newborn screening initiatives, including two upcoming publications, and our annual newborn training boot camp.

The EveryLife Foundation is dedicated to leading evidence-based policy efforts. As the Committee may be aware, in 2021 the National Economic Program for Rare Disease study, in 2022 published those findings for that. The test study included the largest survey of the rare disease community to date, which revealed an average diagnosed odyssey of over 6 years and 17 specialists and providers visited.

Over the last 18 months, EveryLife has been working with the Lewin Group to better understand the economic impact of the diagnosed odyssey in rare disease, and any economic implications for early intervention. Review showed the findings for the new study, cost of delayed diagnosis in rare disease, the health economic study in September.

Of note, the study includes an assessment of the economic impact of the diagnosed odyssey in five pediatric conditions, including SKID, ALD, Pompe, Duchenne muscular dystrophy, and Fragile X. We are also happy to share that during the coming weeks we will be posting a white paper reflecting policies that were proposed as part of a newborn screening by this roundtable series last summer.

The roundtable series brought together more than 100 leaders from a broad array of newborn screen

stakeholders, including academic researchers, state public health officials, patient advocacy organizations and industry and government officials. Together we worked to identify key actual recommendations designed to achieve newborn screening modernization.

Our hope is that these actions will help to transform the newborn screening system to remain one of the most accessible of health programs. We look forward to working alongside you, as we further develop these policy concepts.

And now, a special save the date. An invitation. We are delighted to share that the EveryLife Foundation is once again partnering with Expecting Health for our fifth annual newborn screening bootcamp. Bootcamp will take place in person in Rockville, on Wednesday, November 1, just prior to the final fall Advisory Committee meeting. Our goal is to provide attendees with the opportunity to learn and discuss developments in newborn screening of experts and patient advocates, currently navigating the newborn screening process.

As we also work to facilitate engagement across our ecosystems, we appreciate the time and education of the expert speakers, and community members who will be part of this event. Thank you for the opportunity to

speak to the Committee today, and your tireless efforts on behalf of the nation's newborn screening families. Thank you, and have a great rest of your day.

DR. CALONGE: Thank you, Dylan, And thanks to all of the folks who have provided public comment today, and of course those who shared with us yesterday. And we appreciate, especially parents, bringing their stories forward in such a way that things are work in life, with real examples of impact, these conditions on families and siblings and others.

So I thank you for your time today. We do appreciate it. We listen intently and it's a very important part of our process on this Committee.

At this time, I'd like to say that we'll take a 20-minute break. We're a bit ahead of schedule by like 3 minutes, I think, so we're doing well. And we'll come back in about 20 minutes. Thank you.

(Break)

Expedited Review Discussion

DR. CALONGE: Thanks folks. I think what I'd like to do is just go through the revised slide sets on the expedited review process, amended after yesterday's discussion, and we'll move on with the rest of the agenda. So could we bring those slides up?

And you remember we talked about the background

that there might be a short timeframe that would be sufficient to address issues of the nomination. That was voted against by the Committee. Next slide.

And at this point the nominators might choose to respond to address issues and submit new evidence or other revisions within the one-year timeframe. We believe the ERG could do an expedited review in an efficient manner, incorporating new evidence or just addressing other revisions without starting over. Next slide.

So this is the process. Step one, the Chair sends a letter to the nominators summarizing the issues leading to the decision, and the current practice is within two months of the ACHDNC meeting where the decision was made. That starts the clock on the one-year timeframe, but in that timeframe nominators may resubmit a renomination package for an expedited review. Next slide.

Requests for an expedited review must include responses to the Chair's letter, and may include additional new evidence or information on other relevant issues. The request must outline at least one material change, and include supporting data or documents.

A material change involves a change in scope of the condition nominated, or substantial new evidence for

the nominated condition. If there is a change in scope, it's preferable that there is also new evidence provided in support of the nomination. Next slide.

So these are some caveats that I think we developed after yesterday's discussion. The package must address Committee's questions and comments in the Chair's letter, but nominators must realize that the Chair's letter only includes those issues that were raised in the discussion. There may be other reasons a Committee member voted against adding the condition. So we don't want to create expectations that addressing the issues in the letter alone will result in a changed vote for any one member. Next slide.

The Chair reviews the renomination package and then as necessary can reach out to both the ERG and the Nomination and Prioritization workgroup, and then make the determination if it qualifies as a material change. This step is likely to involve ongoing discussions with the nominators, and will be performed as expeditiously as is practical for all participants.

And we phrased this this way without a timeframe because it really depends on setting up meetings that meet the timeframes and availability of all the parties that are involved, and we're worried if we define this, or shorten it more succinctly, it could

actually work against the process, and kind of create unforced errors. Next slide please.

If the Chair concludes the renomination constitutes a material change, the package will be presented and discussed by the full Committee for consideration for an expedited review. The Committee should vote on whether to move the condition to an expedited review to be conducted by the ERG.

If the vote fails, the Chair will summarize the issues leading to the decision in a letter to the nomination group, and the condition will return to the list of conditions for future nomination and prioritization. Next slide please.

If the Chair concludes there is not a material change, the nominators and the rest of the Committee will be notified, and the condition will return to the list of conditions for future nomination and prioritization. Next slide.

If the vote passes, if necessary, the N&P workgroup will prioritize the review, considering other topics in the prioritization queue, in order to determine timelines and deadlines. The ERG will follow standard systematic review processes to identify relevant research published since completing the previous review.

The ERG may find additional new research on issues not identified in the original review and Committee decision and Chair's letter that could impact the decision to recommend the condition be added to the RUSP. So that would be following kind of standard processes. Next slide.

The ERG will work with the technical evaluation panel, revise the review to include the new evidence, and address the new revision in scope, and this could involve additional modeling. When the review is complete the condition will be scheduled for presentation, discussion and vote, for recommendation for inclusion on the RUSP at a regular Committee meeting.

A vote on the condition must be held within 9 months after approval of the expedited review. Next slide.

It must be clear to all that a topic approved for expedited review may still not be recommended for addition to the RUSP. Next is the intent and expectation of the Committee that this process will be engaged rarely, for very few nominated conditions. This is not intended to become a routine process for most, or even many, conditions.

And finally, recognizing that we need some

experience with this, the Committee review and evaluate the process after two expedited review requests to identify and implement any appropriate revisions.

So I think I've captured the thoughts of the discussion yesterday that you conditionally approved, and hopefully we can have if -- well, let me just open it up for questions and comments. Christine?

DR. DORLEY: Yes, Ned. You did a great job of summarizing everything we discussed yesterday. I just had one question. So if you, as the Chair, or future ACHDNC Chairs, decided there is not an adequate material change, or there's no change that exists, does that nullify the nominator from having a future expedited review for their package?

DR. CALONGE: Yes. So the idea was we had to put it -- I felt strongly we had to put a closure on this, and there is an ongoing dialogue that can last up to a year. But at some point there needs to be a decision that there's not a material change. So there's no timeframe associated with that, but there ultimately needs to be a closure of the door that says we're not going to move ahead with the expedited review.

Part of that is to protect our time, and the time of staff here, and to be clear with the nominators the conversation is not getting to the endpoint. But

you noticed there was no timeframe on that other than the full year, so. Margie?

DR. REAM: Hi. Thanks. I had a couple of questions. First, related to including new evidence. So, if there's a change of scope, and just like a narrowing of the scope, like I anticipate we'll hear later about Krabbe disease, that wouldn't necessarily require new evidence just a change in how the previously reviewed evidence is being applied to the nomination.

So I was wondering if you could clarify the idea of having new evidence with the expedited review request. That's one question. I don't know if you wanted me to give you all the questions, or just do them one at a time?

DR. CALONGE: Let me do that one quickly. So yeah, the idea is that it needs to fulfill one of the two criteria and new evidence for the original nomination, or a change in scope that's a material change. I did put in there that ideally you did both. So you changed the scope, and were able to provide additional evidence, but that's not a requirement.

DR. REAM: Okay. So that's kind of an and/or?

DR. CALONGE: Yes.

DR. REAM: Okay. And then the 9 month, you know, different parts in the Word document that was sent

out, the timeline is more vague than 9 months at different parts in the document. So the 9 months starts from the day of the meeting vote, rather than like how does the decision -- how does the 9 months relate to where something would fall in a prioritization queue?

DR. CALONGE: Yeah. The 9 months is after the decision to do an expedited review.

DR. REAM: Okay. And so I guess how it falls in the prioritization queue could shorten that 9 months, or it could extend it to the full 9 months, but not past the 9 months?

DR. CALONGE: Yes.

DR. REAM: Okay. And then last question -- thank you for taking all my questions. I was wondering if you could comment on what sort of datapoints might be important in the review and re-evaluation of this process once there's two expedited reviews completed?

DR. CALONGE: Well, I think if both were able to meet the timelines. Are the timelines not wrong? Do they need to be readjusted, give the nominators a chance to provide input as to how the process went for them, and see if there are any additional changes that would make it work better?

I think we were also -- we want to make sure

that we are resourced appropriately in terms of staff and time, and Chair time to make this work, and so figuring that out will be important datapoints to say do we need to make any changes.

I would also say that anything else that came up as we know on, I think recognizing that processes are, especially new ones, need to be piloted and tested. I felt strongly that we should do that for this one, and that two sounded to me like a good number to kind of take a relook at how things should be changed, and how we could tighten it up, or sorry, make it more consumer-friendly, or customer-focused, and still keep it within the resource allocation that those of us on the Committee, and those of us who work for HRSA that have available, so thanks for those questions. Shawn?

DR. REAM: Thank you.

DR. MCCANDLESS: Thank you. Shawn McCandless, Committee member. I think more of the questions are very pertinent, but also kind of directed. They sort of emphasize a concern that I've had about this, which is that, you know, I think it's right for this Committee to try and formalize processes and make the process more clear, and make expectations more clear for nominators.

But at the same time I want to understand what is the basis for these like this decision we make today.

What are we creating? Is this a guideline? Is this a part of our bylaws for the Committee? Is this committing HRSA to something? I guess I'm not -- I want to be sure I understand what the level of expectation is because I think that pertinent to some of the comments that Dean Suhr made earlier, we may bump up against problems with timelines in the future if we create very strict policies about what we're going to do this within this timeline.

And so I just want to make sure that I understand what we are voting to create here, what -- how it impacts us, and what are the mechanisms for adapting or adjusting if need be.

DR. CALONGE: Yeah. I appreciate that, Shawn, and I'm trying to build in the flexibility. When we first put it out, other than the one year, there were no timeframes, and that was really to allow us to get some experience with it. Then there was a concern that this process, which was really intended to expedite things, might be inherently slow.

And so, the 9 months the timeline was borrowed from, what the practice we currently follow for conditions that are referred to the ERG for full evidence review, so that's where that number came from. I think the one, the area of flexibility we gave

ourselves was there has to be a vote. It didn't say that we had to vote up or down on the condition at that 9 month period of time.

The vote may be to extend the time for later, and trying to build flexibility in anticipation of the problems that might arise. And then ultimately, I think we need to get some experience, and figure out if this is a process we can do, or can't do, given the resources we have available.

Again, if there's some concerns expressed that this would just be the way that groups do things, and in fact, might have nominators come to the N&P workgroup earlier before the research was robust enough to support a nomination with the idea that they could always come back with an expedited review.

And that's why trying to set the expectations that this is rare, this is not a process for all topics is an expectation I tried to lay out in the caveats for the process. Shawn?

DR. MCCANDLESS: Thanks. And just to clarify, this is not -- is this binding in any way on the Committee? Is this forcing us into, or is this merely an attempt to clarify and document how we intend to process -- a process that we intend to try to follow until we change the process? I guess, is there any

formal meaning to us adopting this as a process? And maybe this is a HRSA question for Jeff Brosco.

DR. CALONGE: Let me just say it won't show up in the bylaws, because our processes aren't in the bylaws, so that's kind of a different issue. But if we had a procedure manual, this would be a procedure manual. We kind of have that with N&P process in the pathways and the timelines, and the other documents that support creation of a nomination package.

I'll let Jeff answer, and then I'll give you my thoughts.

DR. BROSCO: And what Ned said is exactly right. And so, what binds us are things like legislation, and if the Committee decides to take a certain practice, the Committee can decide to change, and it will be, you know, there are bylaws and standard operating procedures, and this falls towards the standard operating procedures, but it's an initial kind of what we think we should do, and let's see how it goes kind of.

DR. CALONGE: Part of the thing, Shawn, with the process now on putting a burden on future Chairs, because there's a lot of decision making and work inherent in the work of the Chair. And I actually think that's appropriate as trying to figure out a way for

someone working directly with the staff could think about the process and to be the point person.

But you recognize there's like words like should vote. But the Committee could choose not to vote on an expedited review process. And so I didn't want to bind the Committee to say the Chair thought this was a material change, it's bringing it forward, therefore we have to vote on it.

I think making that should are ways to say that we're creating this pathway for exceptional topics, not that all topics aren't exceptional, but that it's something about the topic or the evidence, or the evidence review that makes offering this approach something that keeps us responsive to the needs of our advocacy communities. Michele?

DR. CAGGANA: Michele Caggana, member. I guess I'm not clear on how long it takes for HRSA to do the cursory review when a package comes in in the first place. Is that, you know, to kind of fend off the worry that packages will come in sort of incomplete, or really prepared and cause undue burden downstream?

So yeah, I was just wondering, is it pretty obvious if a package, somebody tried to channel something in that direction might -- would be obvious to the staff, and not really be a big burden for everyone

along the line.

DR. CALONGE: The question is -- yeah, go ahead.

DR. BROSCO: Michele, you mean a renomination package, right?

DR. CAGGANA: Yeah. I mean, if something comes in, so there's this fear that this expedited process is going to be used because people are going to come in at the beginning and try and circumvent the whole process, but I think the rules we set in place for this preclude that. But even so, if something comes back with a material change, or extra evidence that's not strong, is that -- unclear, I guess, how much of a burden that will be.

DR. BROSCO: Looking back historically, right, over the last decade. It is very rare for a condition not to be voted to the RUSP once it's past the N&P workgroup. So the initial N&P workgroup has frequently said, no, there's not enough to move forward here, and sent things back. It's happened probably at least five or maybe as many as 10 times over the last decade.

So you're right, there is that initial sort of leap that you have to get to just to get to the Committee to vote for evidence review. What we're talking about now is once you've made it through all the

way there's been a negative vote. And then what I'll tell you is there's a lot of back and forth between nominators, the Chair and the staff, to try to get to a point where we say, yes, there really is new evidence here, there really is a change. This is something that the Committee is likely to take seriously. It's going to move forward.

And the N&P workgroup and the ERG play a role in that as well. So, if the nominators say, well, we have all these new articles, well, we can share them with the ERG, and they could say yes, this really is new, or we both knew this, it's not really new.

And the N&P workgroup can be very valuable because they can say, yeah, this change in scope really is a change in scope. This would change the way we think about things, or, no, it's not really. We also wanted to make it easy because if a study came out that said there's a brand new therapy, it fixes everything, the Chair could just say, yeah, that's a material change.

We don't have to go through ERG or N&P, so there's flexibility built in to meet the many different possibilities of why a condition might come back for an expedited review.

DR. CAGGANA: Okay. Thank you.

DR. CALONGE: Ash?

DR. LAL: I'm actually in support of making the -- narrowing down the reasons why the reconsideration would be granted, or an expedited review would be granted. And I'm especially -- I think the change in scope on material change, that is one point, but the generation of new evidence within a year of the previous deliberation seems like that wouldn't be a frequent occurrence in my view.

Like I think those are the kinds of situations where it could have been better because research from the planning stage to the actual results takes multiple years, and most people would feel aware of results that would be coming down the line. It's better in those situations to just hold off on the nomination to a more favorable time upon expecting positive results, rather than submitting it.

I would certainly I think, oppose the situations when you would expect would be extraordinarily rare, and that it's more often that the other side would be more likely to happen.

DR. CALONGE: Right, and I agree with you. Unless there are objections to what is in front of you being voted on initially yesterday, and I hope to address the issues that came up.

Committee Discussion on Possible Krabbe Expedited Review

DR. CALONGE: What I would like to do is move ahead to present for your consideration a possible expedited review for Krabbe disease, but this would be our first one. There is the summary of Krabbe disease renomination package. Next slide please.

We requested additional published data on the following topics: Evidence review, efficacy of transplants for infantile Krabbe disease that systematically reports outcomes and adequately classifies cases as early vs. late onset, e.g., psychosine levels or genetic markers of infants transplanted.

We requested evidence regarding harms of neuro transplants for infants transplanted in the first two months of life, including the possibility of transplanting infants not meeting criteria for early infantile disease.

And finally, evidence regarding outcomes of infants who are at risk of late infantile Krabbe disease, as these children may experience even greater benefits than early infantile Krabbe disease, and some of those should include number lost to follow-up, and potential burden on families and infants of intensive follow-up visits and consequences of indeterminate

diagnostic testing. Next slide.

Again, material change has to be a change in scope of the condition nominated or new evidence. Resubmission should address Committee questions and comments described in the Chair letter. Next slide.

You remember we voted not to recommend Krabbe for inclusion on the RUSP at our February 2023 meeting. We sent the Chair letter to the Krabbe nominators in March. The disease nominators resubmitted the nomination package in discussions with myself and staff at HRSA in response to the Chair letter, and that came back in July of this year. Next slide please.

I will just point out that we asked for the renomination package in anticipation of adopting an expedited review process. Had we not adopted the expedited review process, we would not have presented the renomination package. So, I want to be clear that in an attempt to be timely with the nominators of Krabbe, we kind of staged this intentionally, and we'd be doing things differently and would have ended the meeting early had we not approved the process.

So, asking the question, has it been less than a year since the ACHDNC vote? The answer is yes. Is there a material change in the new nomination? My assessment of this is yes. The material change was the

change in the targeted screen to early infantile Krabbe disease, defined as onset within the first year of life with reduced GALC enzyme activity, and elevated psychosine in the newborn dried-blood spot, represented a material change in the nomination.

There were additional data provided that we reviewed with the ERG, made the conclusion that the additional data did not rise to the level of a material change, but the targeted screening could change the benefit to harm ratio, which is why I'm asking you to consider this today. Next slide.

Those are the all items, nice and short. Some of the decisions on my part is material change, and now I will put it up for discussion remembering that the next step would be the Committee considering whether to take a vote, and then I would ask if you do phrase the vote in the positive that we would move ahead with expedited review.

And I'll open it up to discussion starting with Committee members and organizational representatives. I often say that one of the physician's best tools is silence.

Vote on Whether to Move Krabbe to Expedited Review

DR. CALONGE: However, I'm going to see if there's someone who is interested in making a motion.

If there is no motion, then we won't move on the consideration of this renomination. Ash?

DR. LAL: Well, I move the motion to for expedited review.

DR. CALONGE: So is there a second?

DR. CAGGANA: I second.

DR. CALONGE: It's been moved and seconded that we move Krabbe disease to the expedited review process based on a material change in scope of the condition. I will now open it up to discussion on the motion.

Seeing no discussion, I would like to move to a roll call vote.

COMMANDER MANNING: Thank you. And if you can just state yes if you are in agreement, no if you're not, or you can let me know if you choose to abstain. Michele Caggana?

DR. CAGGANA: Yes.

COMMANDER MANNING: Jannine Cody?

DR. CODY: Yes.

COMMANDER MANNING: Cynthia Hinton?

DR. HINTON: Yes.

COMMANDER MANNING: Christine Dorley?

DR. DORLEY: Yes.

COMMANDER MANNING: Paula Caposino?

DR. CAPOSINO: Yes.

COMMANDER MANNING: Jennifer Kwon?

DR. KWON: Yes.

COMMANDER MANNING: Ash Lal?

DR. LAL: Yes.

COMMANDER MANNING: Shawn McCandless?

DR. MCCANDLESS: I'm going to take the prerogative of the Committee member to make a comment as I give my vote. And that is that I am going to vote yes that this move forward. I do think this is a substantial and marked improvement in the definition of what newborn screening would look for.

I would appreciate going forward if it were clear that this Committee is made up of people who are capable of understanding evidence and data, and that the Evidence Review Group and the experts that work with them make a good-faith effort to respond to questions and concerns, and I specifically would say two things.

The question about our understanding of whether the addition of psychosine reduces false positives. There's no doubt about that. But to continue saying that there are no false positives when the data presented show false positives as happened in the evidence review, and it was seen in the new literature, regarding of the reason for the false positive, and regardless of whether that is a one off event.

That's real life. And so, please give us the benefit of being intelligent people, and recognizing that there may be false positives. No one has ever asked that there be zero false positives for a condition. This Committee has asked that false positives be balanced with the benefits of the treatment.

The second point I would make is that, again, we all look at the same literature. And when questions are raised about asking for confirmation that individuals that are referred to as having good outcomes from a treatment that questions that are raised to show the evidence that those children truly have the condition up for discussion, and specifically related to Krabbe to refer to older literature where molecular data are not available, where the diagnostic criteria that document the early infantile onset are not available.

It's fine for someone to say, well, we know that these patients had it, but this Committee is not expected to act on hearsay or on the advice of individual experts, and nobody would want us to do that. The data are the data. And if there is a request for data, and a request for additional data, it would be greatly appreciated if that were honored, and honored in a respectful way.

So, with those two comments in mind, I vote yes to move this to expedited review.

COMMANDER MANNING: Thank you, Shawn. Kamila Mistry?

DR. MISTRY: Yes.

COMMANDER MANNING: Melissa Parisi?

DR. PARISI: Yes.

COMMANDER MANNING: Chanika Phornphutkul?

DR. PHORNPHUTKUL: Yes.

COMMANDER MANNING: Jeff Brosco?

DR. BROSCO: Yes.

COMMANDER MANNING: And Ned Calonge?

DR. CALONGE: Yes. Thank you. The motion passes unanimously. Again, to clarify expectations, the strength of the vote does not indicate how we will vote based on the evidence review and presentation, which the clock now starts running on the 9 months.

DR. BROSCO: And Ned, can I bring something up?

DR. CALONGE: Yeah.

DR. BROSCO: Because I just got a text, and I want to be absolutely clear. Organizational reps were allowed to speak. There wasn't any exclusion of organizational reps, so if someone had anything they wanted to say, please, this was not a -- I'm not sure why anyone would think you would be excluded, but no one

was excluded. Okay. Thank you.

DR. CALONGE: Thanks for helping me, Jeff. When I see no hands on the screen I take that as no hands on the screen. At this point, I'd like to ask -- also this will be referred to the ERG. We will use ERG methodology, including a technical expert panel and the Nomination and Prioritization workgroup review as possible, and as feasible, and move forward in time.

New Business

DR. CALONGE: So, at this point I'd like to ask if there's any new business to bring up in front of the Committee? I have Jannine Cody with a hand up.

DR. CODY: Yes. Jannine Cody, Committee member. I was wondering if it might not be a good idea to have a presentation from the -- someone associated with the National Academy of Medicine Project to look at newborn screening, and genome sequencing. And I don't know what their goals are, what their scope is, and I thought it might be, if others agree, a good idea to hear from someone associated with that project.

DR. CALONGE: Yeah. I think, Jannine, in my understanding that it's still early in the negotiation process, so while the money has been identified in the budget, it's still in the process of talking about what the statement of tasks will be. The National Academy's

work is dependent on that negotiated statement of tasks, which comes from a dialogue with the sponsoring organization and the leadership on the board to which the topic has been assigned.

And so, that will occur. Once the statement of tasks is formed, and they start to work, there is no reporting out. It is a confidential process, and it remains confidential up until the end. Now, that being said, the Committee -- National Academy almost always includes public comment, and open sessions to inform their work.

When I did the PFAS study we did town halls. We actually made sure we gained the voice of the affected community across the country. And it would be my hope that as we set the new standard for community engagement for nation's studies with that study, that that would be also a process that was included in this one coming up.

So, I think we could have a presentation, but there are other things that have to occur before that would be possible. So thanks. And I'm not involved in those negotiations, so I just know about them from my own experience with them. Natasha?

MS. BONHOMME: Hi. Natasha Bonhomme, Genetic Alliance. Now two things that Jannine's comments made

me think of. I just wanted to update the Committee that on June 7, the National Academies did have their workshop on Next Generation screening.

DR. CALONGE: You were a co-chair?

MS. BONHOMME: Yep. The promise and perils of DNA sequencing of newborns, and I just wanted to let people know that the proceedings for that are up on the website. I did say that I would update this Committee, and there will be a writeup on that.

I'm happy to share any more information for anyone who would be interested in that. And then the second piece is, you know, this concept of harms keeps coming up, and I know that we've had some sessions that have kind of talked about it. But I wonder if in future meetings we could delve more into that.

I'm thinking of earlier today kind of the message from our speaker, you know, to call a thing a thing. You know, let's put names to things, and it may be helpful to really revisit what does that mean and have some conversation both about harms in newborn screening as well as benefits, since those keep coming up, but not necessarily the details behind those words.

So, just a suggestion as it's a theme that, as I said, keeps coming up. Thanks.

DR. CALONGE: I appreciate that, Natasha. And

I think that is one of the topics that we hope to pursue with a broader inclusion of families and advocacy groups and experts, as we think about a more detailed operationalization of decision making supported by the matrix.

So, we've been talking around it, and I think as I'm talking with Jeff and HRSA staff, purposefully thinking about how we quantify, describe both harms and benefits moving forward to support decision making is something we're all very interested in.

MS. BONHOMME: Yeah. And also just I'm not saying that you were saying this, but I think it isn't just a theme that just advocates or families need to understand, but we, as a whole community and system, need to understand where each of us, how we see that and can work together around those concepts, so thank you.

DR. CALONGE: I appreciate that. And I wasn't saying just families. I think I didn't say just families.

MS. BONHOMME: That's what I said, that's why I wanted to echo that. So thanks.

DR. CALONGE: Thank you. Jennifer?

DR. KWON: I don't have anything new. I just wanted to thank Natasha for bringing up the important concept of harms, and also for trying to clarify what

Shawn was saying about harms yesterday. I think Natasha, that one of the things that Shawn may have been alluding to is that for certain people and populations, it is really hard for them to share their experience, their negative experiences of newborn screening.

So this may be families who are, you know, probably grateful that their child does not have a condition, and maybe as severe as the condition that advocacy groups are passionate about.

It may also be medical providers who hear things that may present medical liability, and are difficult to bring up because of the need to be, you know, really discreet about comments that may, you know, raise concerns about medical liability, or just things that families do that have nothing to do with the advice they were given, but maybe just a misunderstanding of that advice.

So, it's always been a struggle for me to know where those -- that miscellaneous group of harms can fit in, but I think that they also occur, and they're more likely to occur with very rare diseases where treatments are in the hands of, you know, relatively few experts.

So, I just sort of throw that out there in any sort of discussion of problems. I wonder if there could be room for those sorts of topics as well.

DR. CALONGE: Thanks Jennifer. Scott?

DR. SHONE: Not to belabor the harms point, but I think that Natasha was just suggesting, and the back and forth after her comment reflects that there is a need for this discussion, and it needs to include diverse viewpoints and perspectives.

And so I would -- based on just the feedback that you provided, Ned, and now Jennifer, I think it would be important to have that, and I would second that need for a discussion, particularly in light of some of the recent publications that have come out discussing not only this Committee's, but in general, the weight of benefits and harms, and our entire system would benefit from it, so I second her suggestion.

DR. CALONGE: Thanks Scott. Are there any other comments, or any other new business to be brought in front of the Committee this day? Seeing none, as always, I wish to thank members of the Committee, our organizational reps, other members of the audience, those families and other advocates and experts who bring their time forward to provide public comment.

And then all the people at HRSA without which there would be no Committee, there would be no RUSP. There would be no ongoing work. The people that got me to Rockville successfully, and hopefully will get me

home, and who ride herd on our IT, including all the little glitches that we may have from time to time. I appreciate all the work that people sitting in the room with me do, as well as those who are outside the room, making things happen.

And with that, I would like to adjourn the meeting, but I will tell you our next meeting is in person in Rockville, November 2nd and 3rd, 2023. All of the dates through 2025 are listed on the ACHDNC website. And the May meeting of the Advisory Committee on Heritable Disorders in Newborns and Children is -- sorry, the August meeting is now adjourned.

Thank you.

(Whereupon at 12:57 p.m. the meeting was adjourned.)