The Advisory Committee on Heritable Disorders in Newborns and Children

HRSA Meeting

HRSA HEADQUARTERS

5600 FISHERS LANE

ROCKVILLE, MARYLAND 20857

November 1, 2018

1:30 p.m. - 5:30 p.m.

OLENDER REPORTING, INC.
1100 Connecticut Avenue NW, #810, Washington, DC 20036
Washington: 202-898-1108 • Baltimore: 410-752-3376
Toll Free: 888-445-3376

A P P E A R A N C E S

COMMITTEE MEMBERS:

MEI BAKER, M.D., Professor of Pediatrics, University of Wisconsin School of Medicine and Public Health, Co-Director, Newborn Screening Laboratory, Wisconsin State Laboratory of Hygiene

SUSAN A. BERRY, M.D., Professor and Director, Division of Genetics and Metabolism, Department of Pediatrics and Genetics, Cell Biology & Development, University of Minnesota

JOSEPH BOCCHINI, JR., M.D., (Chairperson), Professor and Chairman, Department of Pediatrics, Louisiana State University

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

CYNTHIA M. POWELL, M.D., Professor of Pediatrics and Genetics, Director, Medical Genetics Residency Program, Pediatric Genetics and Metabolism, The University of North Carolina at Chapel Hill

ANNAMARIE SAARINEN, Co-Founder, CEO, Newborn Foundation

SCOTT M. SHONE, Ph.D., Senior Research Public Health Analyst, RTI International

BETH TARINI, M.D., M.S., FAAP, Associate Director, Center for Translational Science, Children's National Health System

EX-OFFICIO MEMBERS:

CARLA CUTHBERT, Ph.D., Centers for Disease Control and Prevention, National Center for Environmental Health

KELLIE B. KELM, Ph.D., Food and Drug Administration, Division of Chemistry and Toxicology Devices

MELISSA PARISI, M.D., Ph.D., National Institutes

of Health, Eunice Kennedy Shriver National Institute of Child Health and Human Development

JOAN SCOTT, Health Resources and Services Administration, Maternal and Child Health Bureau

KAMILA B. MISTRY, Ph.D., MPH, Agency for Healthcare Research & Quality

DESIGNATED FEDERAL OFFICIAL:

CATHARINE RILEY, Ph.D., MPH, Health Resources and Services Administration, Genetic Services Branch, Maternal and Child Health Bureau

ORGANIZATIONAL REPRESENTATIVES:

NATASHA F. BONHOMME, Genetic Alliance

SIOBHAN DOLAN, M.D., MPH, March of Dimes, Department of Obstetrics & Gynecology and Women's Health, Albert Einstein College of Medicine and Montefiore Medical Center

DEBRA FREEDENBERG, M.D., Ph.D., American Academy of Pediatrics, Texas Department of State Health

Services
CHRISTOPHER KUS, M.D., MPH, Association of State & Territorial Health Officials, New York State Department of Health
SHAWN E. MCCANDLESS, M.D., Society for Inherited Metabolic Disorders, Genetics and Metabolism, Children's Hospital Colorado
JED L. MILLER, M.D., MPH, Association of Maternal & Child Health Programs, Office for Genetics and People with Special Health Care Needs, Maryland Department of Health Prevention & Health Promotion Administration
ROBERT OSTRANDER, M.D., American Academy of Family Physicians, Valley View Family Practice
SUSAN M. TANKSLEY, Ph.D., Association of Public Health Laboratories, Laboratory Operations Unit, Texas Department of State Health Services
CATE WALSH VOCKLEY, MS, CGC, National Society of Genetic Counselors, Division of Medical Genetics, Children's Hospital of

Pittsburgh

MICHAEL S. WATSON, Ph.D., FACMG, American College of Medical Genetics

BRITTON RINK, M.D., M.S., American College of Obstetricians and Gynecologists, Mount Carmel Health Systems

C O N T E N T S

P R O C E E D I N G S

DR. JOSEPH BOCHHINI: Good morning, everyone. I would like to welcome you to the fourth meeting of the Advisory Committee on Heritable Disorders in Newborns and Children for 2018. We will begin the meeting by taking a roll call. Going alphabetically:

DR. JOSEPH BOCCHINI: Mei Baker?

DR. MEI BAKER: Here.

DR. JOSEPH BOCCHINI: Susan Berry?

DR. SUSAN BERRY: Here.

DR. BOCHHINI: I'm here.

DR. JOSEPH BOCCHINI: Jeff Brosco?

DR. JEFF BROSCO: I'm here.

DR. JOSEPH BOCCHINI: Centers for Disease Control and Prevention, Carla Cuthbert?

Dr. CARLA CUTHBERT: Here.

DR. JOSEPH BOCCHINI: Food and Drug Administration, Kelli Kelm?

DR. KELLI KELM: Here.

DR. Bocchini: Agency for Healthcare Research & Quality, Kamila Mistry?

DR. KAMILA Mistry: Here.

DR. JOSEPH BOCCHINI: The National Institutes of Health, Melissa Parisi.

DR. MELISA PARISI: Here.

DR. JOSEPH BOCCHINI: Cynthia Powell.

DR. CYNTHIA POWELL: Here.

DR. JOSEPH BOCCHINI: Annamarie Saarinen.

MS. ANNAMARIE SAARINEN: Here.

DR. JOSEPH BOCCHINI: Health Resources and Services Administration, Joan Scott?

JOAN SCOTT: Here.

DR. JOSEPH BOCCHINI: Scott Shone.

DR. SCOTT M. SHONE: Here.

DR. JOSEPH BOCCHINI: Beth Tarini.

DR. BETH TARINI: Here.

DR. JOSEPH BOCCHINI: And our DFO, Catharine Riley.

DR. CATHARINE RILEY: Here.

DR. JOSEPH BOCCHINI: For our organizational representatives, the American Academy of Family Physicians, Robert Ostrander.

DR. ROBERT OSTRANDER: Here.

DR. JOSEPH BOCCHINI: American Academy of Pediatrics, Debra Freedenberg.

DR. DEBRA FREEDENBERG: Here.

DR. JOSEPH BOCCHINI: American College of Medical Genetics, Michael Watson.

DR. MICHAEL S. WATSON: Here.

DR. JOSEPH BOCCHINI: American College of Obstetricians and Gynecologists, Britton Rink by webcast.

DR. BRITTON RINK: Here.

DR. JOSEPH BOCCHINI: Association of Maternal Child Health Programs, Jed Miller.

DR. JED MILLER: Here.

DR. JOSEPH BOCCHINI: Association of Public Health Laboratories, Susan Tanksley.

DR. SUSAN M. TANKSLEY: Here.

DR. JOSEPH BOCCHINI: Association of State and Territorial Health Officials, Chris Kus, by webcast.

DR. CHRIS KUS: Here.

DR. JOSEPH BOCCHINI: Department Defense, Adam Kanis is unavailable today.

Genetic Alliance, Natasha Bonhomme.

MS. NATASHA F. BONHOMME: Here.

DR. JOSEPH BOCCHINI: March of Dimes, Siobhan Dolan, by webcast.

DR. SIOBHAN DOLAN: Here.

DR. JOSEPH BOCCHINI: National Society of Genetic Counselors, Cate Walsh Vockley.

MS. CATE WALSH VOCKLEY: Here.

DR. JOSEPH BOCCHINI: Society of Inherited Metabolic Disorders, Shawn McCandless.

DR. SHAWN MCCANDLESS: Here.

DR. JOSEPH BOCCHINI: Thank you.

So we're a little different format today based on availability of this room. So the workgroup met first, and I guess since it's afternoon and it's the day after Halloween, we'll sort of watch and see how many of you shared your candy -- or shared your children's candy overnight.

(Laughter)

So first on the agenda is approval of the August minutes. Committee members received the draft of the minutes, of the October meeting to review prior to this meeting. We incorporated revisions submitted by Committee members Committee members and distributed the final draft of the minutes to the Committee prior to this meeting.

Are there any further additions or corrections to be added to the minutes?

Hearing none, we just need to vote on the approval of the minutes. And I don't have an approval sheet. So we'll just go down and -- here we go.

Mei Baker?

DR. MEI BAKER: Approve.

DR. JOSEPH BOCCHINI: Sue Berry?

DR. SUSAN BERRY: Approve.

DR. JOSEPH BOCCHINI: I approve.

Jeff Brosco?

DR. JEFFREY P. BROSCO: Approve.

DR. JOSEPH BOCCHINI: Carla Cuthbert?

DR. CARLA CUTHBERT: I approve.

DR. JOSEPH BOCCHINI: Kellie Kelm?

DR. KELLIE B. KELM: Approve.

DR. JOSEPH BOCCHINI: Kamila Mistry?

DR. KAMILA MISTRY: Approve.

DR. JOSEPH BOCCHINI: Melissa Parisi?

DR. MELISSA PARISI: Approve.

DR. JOSEPH BOCCHINI: Cynthia Powell?

DR. CYNTHIA POWELL: Approve.

DR. JOSEPH BOCCHINI: Annemarie Saarinen?

MS. ANNAMARIE SAARINEN: Approve.

DR. JOSEPH BOCCHINI: Joan Scott?

MS. JOAN SCOTT: Approve.

DR. JOSEPH BOCCHINI: Scott Shone?

DR. SCOTT M. SHONE: Approve.

DR. JOSEPH BOCCHINI: And Beth Tarini?

DR. BETH TARINI: Approve.

DR. JOSEPH BOCCHINI: Thank you. The minutes are approved.

So next slide. And one more

So just to remind everyone, there will soon be an announcement for nominations of new committee members for 2019 and 2020. Self-nominations will be accepted as well as nominations made by individual colleagues.

There will also be a call for new workgroup members. All three workgroups are looking for additional members, all three of our standing workgroups. And so this will be announced very shortly. Information on the application process and the dates that the applications will be due will be posted on the Committee website, will be sent out over a variety of list serves so that we could bring in additional individuals from a wide potential audience.

We also wanted to remind you that we've had a call for additional organization representatives. We will add an additional announcement for organizational representatives.

Currently we have three openings. More information will be, again, available at the Committee website, and we want to thank those organizations that have already expressed interest in providing information. They are a part of the group that will be evaluated.

So just some activities -- update you on activities. As I mentioned in August, we are planning a full review of the condition nominating process and of the evidence review. And we will investigate methodologies for recommending a condition for removal from the RUSP within this process.

We've created the steering committee that we've talked about that will help guide this project. The principles of evidence review, obviously, have evolved over time, and we need to determine whether we need changes to the evidence review process to keep up with those changes in the field.

The review of evidence-based review

process will include how evidence and information are gathered for the evidence review, types of data and information that needs to be included, how the evidence is rated and presented to the Committee, and the appropriate method for determining the strength of the evidence. The matrix used for decision-making process will also be evaluated. Our aim is to update the decision-making framework with the latest approaches used in evidence to successfully develop public health policies.

We also have two new projects. As you know, the Committee has a Congressional mandate to follow timeliness. As such, we will examine the progress that's been made regarding timeliness and newborn screening in the United States, led by Alex Kemper and K.K. Lam. This review will serve as an update to the December 2016 GAO report on the timeliness in newborn screening that covered the original data. This effort will provide up to date analysis of how states are progressing toward the Committee's

timeliness goals. The second is an assessment on the implementation of the more recently added conditions to the RUSP.

Over the next year we'll look into the impact of adding those conditions to the RUSP. We plan to conduct a retrospective analysis of how implementation of screening for these new conditions have gone and the impact they have had on public health programs. We would like to determine if the estimated time frames that were predicted are accurate. The barriers and challenges encountered were what the states anticipated, and whether there were any unexpected challenges.

We also want to take a closer look at the clinical and public health implications of adding conditions with known delayed onset and severity.

And then the last announcement is the development of a new ad hoc workgroup. We discussed this a little bit at the August meeting. So we've now established an ad hoc

workgroup to address the overlapping issues that were identified through our review of risk based on newborn screening test results.

Through the Laboratory Workgroup deliberations, interactions with APHL, and Committee discussion, two significant areas were identified within which there may be a role for Committee involvement. One is with education, and the other is with possible policy conservations for stakeholders, including states and clinicians.

Mei Baker has agreed to serve as Chair of this workgroup. This workgroup brings expertise from the Committee, the Laboratory Workgroup, and the Education and Training Workgroup. They had their first meeting earlier this morning, and we're going to hear from Mei tomorrow afternoon, as some of the initial considerations were brought up at that meeting.

Next slide. This shows our next meeting, the date of the April meeting has changed, just

to remind everybody. The meeting was originally scheduled for April 22nd and 23rd. It will now be held on April 23rd and April 24th.

So our February meeting, which is the next meeting, is going to be in person and by webcast. The April meeting will also be an in-person meeting with webcast.

And then you see listed -- those should be the 2019 dates for August and November. Okay. And then the meeting dates through 2020 can be found on the Committee's website.

So meeting topics for today: We're going to review a nomination that we received on cerebrotendinous xanthomatosis, CTX. We're going to have an update on baby's first test, and a panel discussion of educational activities underway or completed in newborn screening.

Tomorrow we will have a panel discussion on the ethical, legal, and social implications of genomic sequencing in newborn screening, and we'll have a presentation on ethical, legal,

social, and policy considerations for newborn screening pilot studies. And then we'll have the updates from the workgroups.

So now I'd like to turn this over to Catharine to go over DFO slides.

Catharine.

DR. CATHARINE RILEY: Thank you, Dr. Bocchini. And good afternoon to everyone who has joined us here in person and who is joining us via the live-streaming webcast. We are glad that you are joining us this afternoon, and hopefully you'll be joining us again tomorrow.

I just have a few logistics to go over. The Advisory Committee's legislative authority is found in the Newborn Screening Saves Lives Reauthorization Act of 2014. This legislation established the Committee and provides the duties and scope of work for the Committee. However, all the Committee activities are governed by the Federal Advisory Committee Act or FACA, which sets the standards for the establishment,

utilization, and management of all Federal Advisory Committees. As a Committee member on the Federal Advisory Committee, you are subject to the rules and regulations for special government employees.

I want to remind Committee members that as a Committee, we are advisory to the Secretary of Health and Human Services, not the Congress. For anyone associated with the Committee or due to your membership on the Committee, if you receive inquiries about the Committee, please let Dr. Bocchini and myself know prior to committing to an interview.

And I also must remind Committee members that you must recuse yourselves from participation in all particular matters likely to affect the financial interest of any organization with which you serve as an officer, director, trustee, or general partner, unless you are also an employee of the organization, or unless you've received a waiver from HHS authorizing you to participate. When a

vote is scheduled for an activity -- or an activity is proposed and you have a question about a possible conflict of interest, please let me know as soon as possible.

According to FACA, all Committee meetings are open to the public. If the public wish to participate in discussion, the procedures of doing so are published in the Federal Register and are announced at opening of the meeting. For this particular meeting, the public comment is going to be coming up shortly. It's the first agenda item, and we have six individuals who have requested to provide oral public comments today. We also received two written public comments that were provided to the Committee members before the meeting. Any further public participation will be solely at the discretion of the Chair, Dr. Bocchini, or myself.

Just a couple reminders about being in the building: For visitors, all those non-HHS employees, visitors only have access to the

pavilion, cafeteria, restrooms, and meeting areas. So all other areas of the facility are restricted and do require an escort by a HRSA staff member. There are no exceptions to this.

If you do need to leave and reenter, you will be required to go through security again, and you will require a HRSA escort to meet you at the security front main entrance. We will have escorts out there around lunchtime and around breaks to be able to help you with that.

We also wanted to remind everyone that visitors are not allowed to take any video or photography in the building under any circumstances. Also in the case of an emergency, we ask that you please exit through the front doors where you entered this afternoon across the street and meet on the parking pad to the left. There will be escorts available from HRSA in the event of an emergency.

Also in the event of an emergency, our security officials ask to only take essential

items, as to expedite the exit and reentry process.

So those are all of the logistics. I wanted to pause and ask the Committee members if there are any questions.

Nope. Great.

Again, welcome, everyone. We are really excited about this meeting. We look forward to the next two days. So I'll turn it back over to you, Dr. Bocchini.

DR. JOSEPH BOCCHINI: All right. So as Catharine said, next time on the agenda is the public comments. We have six people who have requested to make public comment, and they will be speaking about cerebrotendinous xanthomatosis.

The first to join us today -- and we would like each of you, as you give your presentation, to come up to the microphone -- is Dr. Robert Steiner, clinical professor at the University of Wisconsin.

DR. ROBERT STEINER: Good afternoon.

Chairman Bocchini, Committee members, and all, thank you for the opportunity to address the Committee today. Your Committee has an awesome responsibility, and I applaud you all for your willingness to serve.

My name is Robert Steiner. I'm here as a private citizen and an advocate for improving care for individuals with cerebrotendinous xanthomatosis or CTX for short, patients. I'm a practicing pediatrician, geneticist, and researcher will a longstanding interest in newborn screening, having served as a member of HCMG's expert panel that recommended expanded newborn screening.

I take care of children and adults with CTX, and CTX is a primary are of research for me. In fact, I've cared for some of the patients you'll hear about today. I've been managing patients with CTX for more than two decades.

Now, CTX is a devastating disorder, but it doesn't have to be. There are numerous

publications describing successful treatment for CTX and treatment outcome spanning 40 years since the initial development of chenodeoxycholic acid or CDCA as a treatment by Dr. Gerry Salen.

I've seen the two extremes of CTX. I've seen CTX that's diagnosed quite late after a patient has already suffered complications, including dementia and cataracts. Although in that case treatment was begun when the diagnosis was finally made, CDCA has had little beneficial effect on the child's autism and dementia.

Second, I've seen CTX diagnosed in early childhood, allowing early and prompt institution of treatment. In that case, the child never developed autism or dementia and is healthy and well today as an adult with little evidence of disease, and in fact, has a newborn baby.

CTX can also present quite early in the neonatal period with severe liver disease.

Untreated, CTX is a devastating disease with a high probability of neurologic

progression. Affected patients develop diarrhea, cataracts, loss of cognitive skills, autism, seizures, and ataxia among many other symptoms. CTX, however, is very treatable.

Unfortunately, this is a rare disease with signs and symptoms that overlap with other more common conditions. Most healthcare professionals have never heard of it. These factors contribute to a typical delay in diagnosis of CTX of approximately 10 years. In other words, patients with CTX typically see doctors for 10 years with symptoms before a diagnosis is properly made. This delay in diagnosis is critically important because many of the symptoms can be irreversible.

In addition, the treatment is very safe and effective, if begun early in the disease process. CDCA effectively prevents the complications of CTX.

You might ask: How can we reduce that delay in diagnosis; how it may be possible to

shorten or altogether avoid the diagnostic odyssey? Educational efforts might help, but doctors simply don't have the time to hear about another rare disease that they're not likely to see during their entire career.

I'm thoroughly convinced that the only way we're going to reduce the time to diagnosis in CTX is through newborn screening. Fortunately colleagues, including Dr. Andrea DeBarber, who you will hear from today, have developed methods for newborn screening for CTX using the same filter paper dried blood spots already in use in newborn screening programs. Furthermore, the method uses the same mass spectrometry instruments in use in these programs. It is reliable, accurate, and reproducible.

We have the opportunity to prevent the catastrophic effects of CTX and save additional patients from suffering. All the elements for a successful CTX screening program are in place. A newborn screening method has been developed.

Confirmatory testing, by way of genetic sequencing and measurement of cholestanol is widely available. A forty-year-old safe effective treatment is available, and CDCA is being used to treat nearly 100 US patients currently.

Research studies like one recent US study that screened approximately 170 children with bilateral cataracts for CTX and identified three with CTX make it clear that we will find cases in the US population if we look.

Physicians such as myself with expertise in genetics, metabolic disease, and lipid disorders are available to treat CTX patients.

I implore the Committee to take the next step towards recommending newborn screening for CTX. As a scientist and medical journal editor, I understand that in an ideal world all the I's would be dotted and all the T's crossed prior to such a recommendation. We would love to see large prospective pilot studies in the US carried

out using the new methods for CTX screening with large numbers of newborns successfully identified and treated and the results published. All that, however, takes a great deal of time and money.

The funding for such studies is not immediately available, and time is precious. Every day we delay instituting newborn screening is another day that CTX patients wait for diagnosis and treatment with life-altering irreversible complications developing.

We know very well the biology of CTX. We understand the treatment, and we are available to take care of the patients. Let's begin to help these patients today, please. Thank you.

DR. JOSEPH BOCCHINI: Thank you, Dr. Steiner. And thank you for all your work in this field.

Next is Dr. Andrea DeBarber, research associate professor at Oregon State Health and Science University.

DR. ANDREA DEBARBER: Good afternoon,

Committee members. Thank you for the opportunity to speak today. My name is Andrea DeBarber, and I am lead nominated for the team submitting the nomination package for consideration of CTX for addition to the RUSP.

I am a researcher at Oregon Health and Science University and have been working with a number of collaborators to develop tandem mass spectrometry methodology capable of screening newborn dry blood spots for CTX.

Today I would like to provide an update to the Committee on the study of ongoing pilot studies to screen newborn dry blood spots as well as some recent developments to ensure the methodology to screen newborn dry blood spots would work well across different platforms and in different laboratories.

Newborn dry blood spots can be screened for CTX using a published negative mode tandem mass spec method with flow injection analysis of tetrol glucuronide species, biosorts (phonetic spelling),

and the use of metabolite ratios. The low force positive rate for flow injection analysis can effectively be reduced to zero using tandem mass spec analysis with liquid chromatography.

Methodology that has been demonstrated to work well in my laboratory and that can incorporate analysis of an additional confirmatory market for CTX 7 alpha, 12 alpha, dihydroxy-4-cholesten-3-one.

Using liquid chromatography tandem mass spec analysis of tetrol glucuronide species and biosorts, Dr. Michael Gelb at University of Washington has screened around 30,000 de-identified dried blood spots from Washington State newborns for CTX. Using dried blood spots from CTX patients, he has shown that the tetrol glucuronide marker is well elevated above the cutoff chosen for the study, and that all newborn dry blood spots screened so far fell well below the cutoff with a consequent force positive rate of zero per 30,000. The goal of this pilot study

is to screen more than 100,000 newborn dry blood spots.

In collaboration with Dr. Tzippi Falik Zaccai, my laboratory continues to screen identifiable newborn dry blood spots for CTX from the Northern Israel region where around one in four newborns are members of the high-risk Drusbum (phonetic spelling) community. Our goal is to screen at least 10,000 newborn dried blood spots.

Other researchers are working to make data available from additional pilot studies screening newborns for CTX. Subject to NIH grant funding, colleagues in the US are planning to perform a perspective pilot study beginning 2019 that would screen more than 135,000 dried blood spots obtained from identifiable newborns born in New York State for CTX.

In the Netherlands where the Dutch Health Council has advised adding CTX to the Dutch Newborn Screening Program upon availability of a suitable screening method, colleagues are

planning to perform a prospective pilot study beginning 2019 with the goal to screen around 20,000 dried blood spots from Dutch newborns for CTX.

Recently my laboratory has confirmed the identity of the primary tetrol glucuronide disease marker present in CTX newborn dry blood spots by comparison with custom synthesized authentic standard. The standard in stable isotope labeled internal standard has been made available to the CDC's newborn screening and molecular biology branch to aid in development of an external quality assurance system to screen newborn dry blood spots for CTX, and efforts are underway to perform between laboratory methodology comparison studies.

In summary, we have made great progress over the last few years to develop and validate newborn screening methodology for CTX that is sensitive specific and that has been demonstrated to work well in a number of different

laboratories.

As Dr. Steiner indicated, we don't yet have data available from large perspective pilot studies screening identifiable newborns for CTX, but we are working to make additional pilot study data available. Every day we delay instituting newborn screening for CTX additional cases are missed with the high likelihood they would not be diagnosed until much later in life when the neurological damage cannot be reversed with treatment.

I appeal to the Committee to move the CTX nomination forward in the consideration process to add disorders to the RUSP. And thank you again for the opportunity to speak and for your deliberation of the CTX nomination.

DR. JOSEPH BOCCHINI: Thank you very much, Dr. DeBarber.

Next we have John Wolf, board member of the United Leukodystrophy Foundation and the father of a child affected with CTX.

MR. JOHN WOLF: Good afternoon. I would like to thank the Committee for giving me the opportunity to speak today.

I'm here today as the father of a CTX affected daughter and as a patient resource and advocate. I'm also the CTX patient liaison for the United Leukodystrophy Foundation.

My daughter Ashley was diagnosed with CTX 14 years ago at the age of 10. She exhibited signs of CTX from birth, starting with chronic diarrhea.

Caring for her as an infant and toddler with this presented a lot of challenges. Leaving the house with her was inconvenient and required deliberate planning around having a bathroom accessible nearby. For example, we would be in line at an amusement park and would often have to leave the line to take her to the bathroom. We would have to plan a trip through security at the airport, not knowing how long we would be in that line. These are things that most people take for granted.

This was especially challenging when traveling and for her while she was in school and any functions away from home.

Over the course of several years and after seeing several specialists for this, no definitive diagnosis was given except irritable bowel syndrome. We were sent home with a prescription for a giant bottle of antidiarrheal medication, which worked or a short while but eventually stopped working entirely, despite increasing the dosage.

It was about halfway through her first grade school year that we were notified by her teacher, who raised the concern of Ashley having issues with paying attention in school and difficulty retaining what she had learned. This was especially puzzling because it was in stark contrast to her development as a toddler, where she had hit all of her milestones early. We had difficulty accepting this, but eventually we began to see and acknowledge it as well and

realized that this was a new reality for her.

In trying to understand what was transpiring with her, we took her to see a psychologist, who ran extensive tests, however, again, no definitive diagnosis was given nor treatment or therapy prescribed. We just adjusted to the fact that she would need additional help in school.

This was status quo until December of 2004 when we noticed that when reading she would hold the book much closer to be able to see it and was also sitting much closer to the TV than before. This was especially alarming, as at the beginning of the school year her vision had tested showing she had 20/20 vision. We very quickly took her in for an examination at a local optometry shop, and we were told that her vision was now 20/100 and that she had cataracts in both of her eyes at the age of 10.

To get confirmation of this, we took her to see an ophthalmologist where a brief history

was taken, and we learned that she did not fit the profile for any of the common causes of cataracts in children. We were very fortunate, however, in that her ophthalmology was curious enough to do some research, which led him to suggest that she could have a metabolic disorder and immediately referred us to a genetic specialist. It was within two months of that referral that we received a confirmed diagnosis of CTX.

Hearing my story, which is very much the abridged version, you might say that sounds like a lot to go through. It absolutely was, but also know that we were extremely fortunate. Most CTX diagnoses are not as easy, and our story is very much the exception to the rule.

She started treatment immediately, and over the course of the first year she began to do markedly better in school and gain 70 pounds, which finally put her into a normal weight percentile.

Now at 24, she lives independently, works, drives, and recently gave birth to my first grandchild. Without her diagnosis at 10, I'm afraid that I would be telling a very different story.

Over the last 14 years, despite efforts to raise awareness in the various medical disciplines that might see a CTX patient, despite the ready availability of a simple blood test to provide an initial diagnosis, despite the isolation of the gene mutation that causes CTX, and despite clearly defined symptoms, and due to it affecting multiple systems in the body, which cause it to clinically present as other more common conditions, this can often distract medical professionals, even when they themselves have CTX-affected children. I know this because this family is in a patient community. All of this can lead them to a dead end and significantly contributes to the average age of diagnosis being the early 30s, inevitably leading

to a devastating outcome for the patient.

To add CTX to the RUSP would all but eliminate the experiences that have affected individuals and their families have had to endure. I can think of no stronger argument to move forward with the CTX nomination and implore you to do so. Thank you.

DR. JOSEPH BOCCHINI: Mr. Wolf, thank you for sharing your family's personal story.

Next we have Kent Richter, who is the spouse of an individual affected by CTX.

MR. KENT RICHTER: Distinguished ladies and gentlemen, good afternoon. I appreciate you allowing me to speak, and I also appreciate the fact that you have the desire to want to help future generations.

So I'm Kent Richter, and that sleeper over there is my wife Donna. Hi, Donna.

MS. DONNA RICHTER: Hi.

MR. KENT RICHTER: And she has CTX. And we traveled from Tavares, Florida, to talk to

you.

Anyway, she's always been physical. I've known her since 17, so more physical than mental, you know. So she used to like to be touched. She doesn't like that now, but she still loves to touch a lot of things.

So I saw pictures of when she was maybe 11 at most, and she already had the same the bumps, we just call them bumps, which wasn't diagnosed, but we know now. And at 24 -- we married at 20, and at 24 she had the larger one taken out. The doctor said, "This isn't what I expected." He says, "This is actually growing out of the tendon that extends there." He says, "I didn't expect that." So he says, "I basically just shaved it. I didn't want to cut into the tendon."

So we were jogging in the 20s -- when we were in our 20s and 30s, and she kept having tripping problems; but we did it. And then one days she says, "I don't want to go jogging now." And she couldn't keep up anyway. She wasn't

gaining any speed. She wasn't getting any stamina or anything, you know. I would down a block and run back, you know, to her and so on. She didn't want to go, okay.

So then I realized -- I mean, her Achilles tendons were huge. I mean, they're this wide. I mean, if you want to see them, we're here. So but they're this wide. So now she's got the bumps here, which this one grew back that was operated on, and the Achilles are wide. Still no diagnosis, going to different doctors.

So then she started having trouble walking, so more than just the tripping. So okay. So we go to -- let's try a neurologist, okay. So the neurologist didn't know. We went back as she was getting worse, didn't know. Then said, well, wait a minute, I know a smart man. It was her teacher, Kenneth Heilman, who was head of neurology at Shadelands at the time, University of Florida.

So anyway, we went and saw him and

diagnosed that visit. Then he sent us to Bill Conner -- Dr. Conner, sorry. She gets to call him Bill and gets to call Dr. Heilman Kenneth. I just -- I need to do it right here.

So anyway -- so that was good. We went out there, you know. He helped us about what to eat, what not to eat, you know, and to try to keep the diarrhea not as bad. It's as bad as John says and having to go to the bathroom a lot.

And so, you know, it was also to help future generations. That was the idea. So basically she has a lack of abilities, you know, a lot of pain, a lot of pain. So thanks.

DR. JOSEPH BOCCHINI: Thank you very much, Mr. Richter.

Next we have Susan Stewart, mother and legal guardian of a young man severely affected by CTX.

MS. SUSAN STEWART: Thank you for having me. My name is Susan Stewart. I am, as he said, the mother and now legal guardian of my son Eric.

Eric is 27 years old, and he has CTX.

Eric was not correctly diagnosed and then appropriately medicated for CTX until he was 16. My pregnancy with Eric and his birth were without incident. I brought home a happy baby, who nursed well and slept through the night at six weeks.

Eric appeared to develop on schedule, and I have no concerns. He seemed on the same developmental schedule as his half-brothers. When Eric was 16 months old his babysitter said, "I have never seen a toddler that likes to play by himself so much." That made me start paying attention to Eric's development more closely. Although he had a few words at 11 months, he lost those words.

At two years I took Eric to his pediatrician because now he had no words and only babbled using vowel sounds. His pediatrician did not seem overly concerned, but he did refer Eric to an audiology who suggested tubes. Although

the tubes solves his ear concerned, it did not solve the developmental concerns.

At two and a half Eric was diagnosed with a communication disorder. At three, he was diagnosed with autism. At five, he was diagnosed with a seizure disorder, Landau-Kleffner syndrome. At six he was diagnosed with a likely immune system disorder, as his MRIs were showing decreased white matter -- sorry -- an increased grey matter in his brain. Around nine, he was identified as intellectually disabled. At 11, he was diagnosed with bilateral cataracts.

During Eric's diagnostic journey I sought the advice of many practitioners, and Eric participated in many therapies at great expense and time for our family. Some of the therapies were scientifically based, but I tried other therapies that were not scientifically based, as I was desperate to help my child.

By the time Eric was 16, he had totally deteriorated from the baby he had been. He was

nonverbal and mainly communicated by typing words, but not sentences, on an augmented communication device. He has never advanced to the sentence level. He can perform -- he could not perform many activities of daily living like bathing, dressing himself, or brushing his own teeth. If we went on an outing of any length, he needed to have a wheelchair. His IQ was 40 on the WISC scale.

Additionally, although he had previously been high energy in his earlier years, he began to have extremely low energy. He just sat on the floor and quietly played with toys. Eric was very ill, and I feared that he was dying.

I did a google search using the words cataracts and autism. I found that disorders related to difficulties with cholesterol -- and believe me, I had been to many doctors, many, many, many doctors. Disorders related to difficulties with cholesterol often had cataracts and autism combined, and I found out that one of

the main research facilities studying these disorders was at Oregon Health and Sciences University.

Eric and I were Oregon residents, so we went to OHSU; and Eric received the correct diagnosis and began treatment. I wonder if I would have discovered his diagnosis if we had lived somewhere else besides Oregon.

Since beginning treatment, Eric has made improvements, but he remains severely disabled and impaired. He is now able to dress himself and buckle his own seatbelt, but he can't cook for himself or bathe himself. He has more energy. His wheelchair has been given away, but Eric continues to have a profound expressive language disorder, a severe receptive language disorder, and moderate to severe autism.

Now I celebrate small achievements. Two years ago, at the age of 25, Eric learned to get himself a cup of water when he was thirsty. Eric currently lives in a group home for

developmentally delayed individuals. He receives Social Security, Medicare, and Medicaid. Additionally, he receives over $9000 a month from the State of Oregon to pay for the caregivers that he needs.

Eric's recent individual service plan was three-fourths of an inch thick with protocols for everything. Eric has no job.

If newborn screening for CTX would have been available for Eric, his life would have certainly had a different outcome. Eric's CTX greatly impacted our family as he grew, and his difficulties continue to impact our family, community, and state.

I hope my children and their grandchildren will benefit from a newborn screener for CTX. And a picture of Eric when he was a baby. Thank you.

DR. JOSEPH BOCCHINI: Thank you, Ms. Stewart, for sharing your family's odyssey. Thank you.

Next we have Robert Rauner, president of the United Leukodystrophy Foundation.

MR. ROBERT RAUNER: Hi. Good afternoon. Thank you to this Committee for taking your time to hear us tell the story that the need for CTX be added to the newborn screening program.

My job, I am the president of the United Leukodystrophy Foundation. I've been a part of the foundation since 1994 when my son Kevin was diagnosed with adrenoleukodystrophy. So I've been a member -- board member of this organization since 2000, and I've been president for the last four years.

One thing I appreciate is work of this group, meaning you, on the work getting X-ALD added to the RUSP back in 2015. As a result of that decision, I became involved with the process of adding X-ALD to the Nebraska Newborn Screening Panel. So I spent two years working through the legislative program to make that happen. So through that time worked with our Newborn

Screening Advisory Committee, and I tried to understand all the processes that happened behind the scenes to bring the recommendations forward to add the disease to our panel.

We began to screen in Nebraska for X-ALD, Pompe and MPS 1 in July of this year, so basically it turned into a three-year process before we made it happen. Throughout the work that I was doing with these diseases, I was asked to become a member with the Nebraska Newborn Screening Advisory Committee. So I've created an extra job for myself, but it was an honor to be asked to be a member of the committee. And so I'm going to do my best to fulfill that role as best I can.

One of the things United Leukodystrophy Foundation has been involved with the CTX community probably since the mid-2000s. It was at that time that I was introduced to John Wolf and his family, and I learned a lot about CTX and had no idea before that time.

In 2009, we at ULF worked with Manchester Pharmaceuticals to help gain orphan drug status for Chenodal as a treatment for CTX. So we at the ULF have been a support group for the CTX community and have supported treatments that have been made available for the benefit of that community. We've partnered up with Retrophin to find ways to work together, not only supporting the Leukodystrophy Committee but especially the CTX families.

We at ULF have also hosted CTX specific meetings, and so that's, over the years, you know, addressing research in the field. And lately our focus has been on Andrea DeBarber's work in newborn screening, and so we've had her and the doctors working together to get through this process as we are today.

One of the things we helped do is facilitate grant funds that CTX community can use to provide families funding to help pay for treatments that may be available.

So as an organization, we will continue to help support the families. We will have meetings and travel grants and other forms of family support.

As a parent, I know how important it is to have the ability to identify what illness your child may be affected with. Newborn screening is a great tool, and it will help families like mine, John's, Sue's, Kent's, and those that are not in attendance today that take the time to send in letters of support.

Having the ability to know, when your child is born, that they may have a disease that can't have a treatment -- if they're identified by newborn screening is a very important thing. What this will do is save them from doing through the diagnostic odyssey that all of our families have gone through.

So I'm very confident in the work that Dr. DeBarber and Dr. Steiner have done leading up to us being here today, and I am confident that

what they've done would be enough for you to accept nomination to add CTX to the RUSP. So thank you for your consideration of adding CTX to the RUSP from all of us here in person and also those that have taken time to give their letters of support. Thank you.

DR. JOSEPH BOCCHINI: Thank you very much, Mr. Rauner.

So that will conclude the public comment portion of this meeting. We will now present a review of the condition nomination for CTX.

So in August the Committee received the nomination package from a team of nominators, led by Dr. DeBarber, and the nomination and prioritization workgroup, which is a subgroup of this Committee, is responsible for performing the initial review to determine if all the elements required for bringing this to the full Committee are present.

The Nomination and Prioritization Workgroup has completed its review, and Dr. Scott

Shone, a Committee member and member of the Nomination Prioritization workgroup will provide a summary of the nominated condition and the workgroup's recommendation to the Committee.

Dr. Shone.

DR. SCOTT M. SHONE: Thank you, Dr. Bocchini, Committee members. Good to see everybody.

As Dr. Bocchini said, I'm going to be presenting the Nomination and Prioritization Workgroup report on cerebrotendinous xanthomatosis, and I will now call it CTX for the rest of the discussion. The Nomination and Prioritization Workgroup includes Dr. Bocchini, Dr. Brosco, Dr. Cuthbert, myself, and Dr. Tarini.

I want to be clear and Dr. Bocchini mentioned this, that the purpose of the workgroup is not to evaluate whether or not there's enough evidence for inclusion of CTX on the RUSP but rather if a nomination package includes all of the components necessary to facilitate or to

suggest that there is enough evidence to permit that review within a nine-month time frame that the External Evidence Review Workgroup must conduct.

So as we've heard, the nomination of CTX was led by Dr. DeBarber, and then was co-sponsored by many additional individuals, some of whom we've heard from today.

CTX is a progressive metabolic leukodystrophy. It is a disease of lipid storage, as we've also heard earlier.

The onset ranges dramatically from birth to adulthood -- and we've heard examples of that from the public comment -- with infantile-onset diarrhea, childhood-onset cataracts, adult to young adult-onset tendons xanthomas as well as deterioration of neurologic function. And then adult-onset progressive neurologic dysfunction from dementia through to seizures.

CTX is an autosomal recessive inheritance but does have, as I said, variable phenotypic

expression. It's a deficiency of mitochondrial enzyme coded by CYP27A1 gene.

But there are more than 57 disease-causing variance found in this gene, and there had been no phenotype genotype correlation with CTX. And the onset and presentation of symptoms is substantially variable with the same pathogenic variant causing different outcomes within even the same family.

It has been reported about 300 CTX patients worldwide over the last 70 years with incidents varying significantly. I listed here 1:130,000 in the South Asian population to 1:470,000 African. There are some where incidence is much higher. Dr. DeBarber mentioned the study in Israel where CTX is much more prevalent, but there is a wide range of instances of the disorder found in the population.

So that's a little background on CTX, summarized what was in the packet that was shared with the Committee.

I want to cover the key questions that the N&P Workgroup considered when reviewing the nomination package.

First, is the nominated condition medically serious?

Is the case definition proposed and the spectrum of conditions well described to help predict both the phenotypic range of the children who will be identified based on the population screening?

Are there prospective pilot data from a population-based assessment available for the disorder?

Does the screening test analytic validity?

Are the characteristics of the screening test reasonable for the current newborn screening system?

Is there a widely available CLIA and/or FDA-approved confirmatory and diagnostic process?

And do the results bear clinical utility?

So if the spectrum of disease is broad, as we've seen here, will the screening and/or diagnostic test identify who is most likely to benefit from the treatments we've discussed and listened to?

And then finally, are there defined treatment protocols available, FDA-approved drugs, and is it widely available?

So I'm going to go one by one through each of these questions.

So the first question: Is the nominated condition medically serious? Yes. CTX is serious. We've heard that from the public comments. The N&P Workgroup felt there was adequate evidence to support that. Despite the range of phenotypes, it's a progressive neurologic disorder. When left untreated, it is very serious when identified clinically, which is obviously the hallmark of a newborn screening disorder, but it is very rare with only 300 cases identified, as I said, in the last 70 years with different incidences -- substantially different incidences based on

the subgroup.

Is the case definition and the spectrum of the conditions well described? The N&P Workgroup felt that, as proposed, the case definition was not well described. They refer to a suspicion index -- a public suspicion index that aids in the clinical diagnosis of CTX. It takes into account a great number of factors, including family history and systemic and neurologic features. And while the most serious phenotype is clear, there is a lack of genotype/phenotype correlation, and minimal data is available on the clinical subtext we see. Also, the case definition in the nomination packet did not have any biochemical markers or profile to aid in identification of CTX cases.

And as been brought up in the oral comments, are prospective pilot data available? And before I get into the answer to that question, which we've heard a little bit about, I wanted to go over the three main features of the

criteria for prospective pilot study data for the Committee to consider the study.

The pilot study proposed should have evaluated the newborn screening process from collection through diagnosis and identify at least one screened positive newborn with confirmation of the condition under the consideration.

The population for the pilot study as well as the screening protocol should be similar to the US population and to state newborn screening programs with respect to known prevalence of the condition, the timing and approach to screening.

And the modality used in the pilot study should be comparable to the method proposed in the nomination application.

And so the answer to this question is also no.

The three pilot studies identified in the nomination packet: The first was the pilot study

in the Netherlands population which had about 200 samples tested. They were all anonymized.

Likewise, the population study that's ongoing in pilot study three listed by Dr. Gelb is an anonymized study -- it's ongoing. There have been no screened positive results yet as well.

There is an identifiable Israeli population study going on currently that Dr. DeBarber mentioned. The caveat to this: The N&P Workgroup felt this challenged the criteria of requiring a population similar to US population with respect to known prevalence. The pilot study focusing on a high prevalence population doesn't accurately affect will we be able to identify this disorder within the system -- within the United States.

Does the screening test have established analytic validity?

This was unclear. This was neither a yes or a no, but based on the information submitted

to the N&P Workgroup, we could not make the determination on analytic validity. There was good data provided, but it was not thorough enough for our evaluation to be completed.

There were two methods suggested, a published paper as well as a manuscript submitted with flow injection, single-tier assay that used the ratios that Dr. DeBarber mentioned as well as a two-tier assay that had much higher specificity.

For both assays, the data submitted was very limited in terms of analytic validity. The manuscript submitted for the two-tier method did not include the supplementary data that was referred to throughout the manuscript. Only accuracy and precision data was provided for between run, not throughout.

Linearity and interference results were discussed but not shown to us. There was no data provided for limits of detection or quantification as well as recovery of the desired analytes.

And matrix effects were indicated. This was a quote in the manuscript: "Matrix effects indicated the need for a stable isotope internal standard," but it was not available at the time of the study presented to the N&P Workgroup.

Likewise, the characteristics of the screening tests proposed were unclear in terms of their reasonableness for the current newborn screening system. The single-tier assay proposed used quarter-inch punch as opposed to the one- eighth-inch punch commonly used throughout the newborn screening system. Or they used two one- eighth-inch punches.

The assays varied substantially in the generation of mass spectrometry used from the Waters Premier XE to the Sciex QTRAP 5500, which spanned the gamut of sort of the lower end mass spec to the higher end mass spec where most newborn screening programs currently use a model that's somewhere in between the two.

The false positive rate was acceptable.

Dr. DeBarber did mention this in her comments, and the false negative rate is unknown at this point because the pilot studies have not adequately been able to assess that, or really it's hard to know this, even with the disorders we screen for currently. So to be perfectly fair, that is a fair unknown at this point.

The assays as presented in the nomination package are not multiplexible with the current analytes on mass spec. There are -- and I have seen -- other methodologies out there. Dr. Gelb has worked on one which is multiplexible, but as presented, the assays were not multiplexible with the current panel as well as the availability and stability of free agents is unknown. CDC is working on this, as is mentioned in your comments, but for widespread use in the newborn screening system, this was an unknown and unclear to us.

And likewise, there will be other disorders detected, which weren't really

effectively discussed other than to say that they would -- they would screen positive for other peroxisome biogenesis disorders, cholestatic liver disease, as well as even Niemann-Pick Type C.

The availability of a clear FDA-approved confirmatory test. The answer to this is most certainly yes. The measurement of elevated cholestanol in blood and elevated bile alcohol glucuronides in urine is well established, the measurement of ketosterol bile acid precursors in blood, as well as genetic testing. And there are multiple CLIA-certified laboratories performing this type of confirmatory test.

It is unclear, however, if the results have clinical utility. Again, this wasn't a yes or a no. But based on the available data for the N&P Workgroup, we felt it was unclear.

There is a broad spectrum of disorder phenotypically with a few cases. The suspicion index I mentioned during the discussion on case definition is a guide for diagnosis. And while

the most serious phenotypes are clear, the progression of other phenotypes is uncertain with very limited data on variance.

Ultimately, it's really unclear how cases would be handled that have a high suspicion but limited clinical findings.

Again, it was mentioned by the speakers before me that there are defined treatment protocols available, and that CDCA treatment is clear though. While low risk, it has some hepatic toxicity and cholic acid has been recommended as a less hepatotoxic treatment but might not be as effective. And the FDA has not granted marketing approval of CDCA for treatment of CTX. However, it was granted orphan drug designation.

So to summarize for the Committee, I listed the key questions again.

The condition is medically serious.

The N&P Workgroup did not feel that the case definition was well described, nor are there available

pilot study data require for the advancement of the nomination.

The analytical validity and characteristics of the screening test were unclear to us.

There is a widely available CLIA FDA-approved confirmatory test.

The clinical utility remains unclear.

But there is treatment available, as has been discussed.

And so the N&P Workgroup found the Advisory Committee will provide guidance to the nominators regarding additional information needed to meet the Advisory Committee requirements to complete the nomination packet as well as additional areas needing clarification. Thank you.

DR. JOSEPH BOCCHINI: Thank you, Scott.

So based on the decision of the Nomination and Prioritization Workgroup, it was felt that we needed to go back to the nominators

and felt that this condition was -- this packet was not ready to be brought to the full Committee for a vote. So there will be no vote today.

I want to compliment the submitters. I think that it's very clear that you have met some of the standards that the Committee requires. In addition, you have some ongoing work that will provide what we hope with the data that's necessary to meet the pilot study requirement that the Committee has. And we think that some of the areas needing clarification could be done through conversation with the Committee and providing the additional data with the work that you're doing.

So we felt overall, this is a very positive submission, but that we still needed more work before we were ready to determine -- before we had all of what was needed to go forward to bring this to the full Committee for a review for potential nomination -- bringing it for evidence review.

So I want to thank you for all you did to get to this point and all the work that you've done to help the families that are affected by CTX.

So with that, let's open this to the Committee to determine if there are any additional questions or comments that they would like to make concerning this submission.

Sue? You probably need to identify yourself.

DR. SUSAN BERRY: There's a sign with a smiley face that says state my name. So this is Sue Berry.

And one of the things that we've consistently observed in bringing new disorders onto the panel is that there are -- and it's going to be clear in this one too, a wide spectrum of presentation. And so even when we have a target for something really important, there are other conditions, not related conditions, but the very condition you're looking

for that are variant. It seems like this is a condition where that's going to be true given what has been described about the phenotypic variability. What I couldn't get a sense of here was how frequently this presents in the neonatal period as opposed to other times.

I think when you screen for things as newborns, once of the targets -- or in many ways we've tried to direct it toward early intervention in the newborn period that needs to be relatively rapid. I understand that there is some benefit of identifying disorders that will have later onset, but knowing the temporal progression and percentage, if you will, that need immediate attention is another question that I didn't get a sense of here. Is that something that can be asked as well, or could you clarify that for us, Scott?

DR. SCOTT M. SHONE: Scott Shone. So in terms of -- correct me if I'm wrong, but I think what you're discussing is sort of the assessment

of benefit of detection through -- yeah. So that's really not the --

DR. SUSAN BERRY: I think it's part.

DR. BETH TARINI: This is like urgency. You have urgency because benefit does not necessarily have --

DR. SUSAN BERRY: I was looking at urgency but also early benefit. Yeah.

DR. SCOTT M. SHONE: I think -- well, what I was going to say still stands. I think that, that's really the point of the external evidence review. I don't think that, that's the purview of the N&P Workgroup. That level of data would come through the next step, and I think that it's okay for that.

So I think -- as I look at Dr. Bocchini. I don't necessarily think it has to be part of the nomination packet. Obviously, it helps guide the next steps or pointing out that, that data does exist. But in terms of the questions that were answered, that level of assessment is not

part of what we looked at or what we look at on a reapplication.

DR. SUSAN BERRY: This is Sue again. I would even settle for a ballpark guess, so that I could sort of put that in my intellectual context as I think about what we learn going forward. But I understand if you don't have it. That's fine.

DR. JOSEPH BOCCHINI: Mei, Dr. Baker.

DR. MEI BAKER: Mei Baker. I want to adding on this a little bit because based on my knowledge for this disorder, it's a little bit different that yours. If we talk about late onset, for example, Pompe, when it's later, and then they have very limited, mild muscle weakness.

But this disorder, what I understand is, actually, the symptoms actually find out early on, just non-specific. People didn't pick it up. Diarrhea - people didn't pick it up. But until you have cognitive function, at that time you

cannot go back.

So we did see an adult patient here, but it's very -- it's very different. I think this pattern is different. So I just wanted to mention that.

DR. JOSEPH BOCCHINI: Dr. Brosco.

DR. JEFFREY P. BROSCO: Yeah. Following up on Mei's point. This is Jeff Brosco. I think that's exactly right. A lot of other conditions we face, the late onset has been so much more mild. It raised questions about, well, how good is it to know early on. This is a case where the late onset is actually -- has devastating consequences very often.

So, yes, you're early detection may be 15, 20 years ahead of time, but it's different in that way. So it really does raise an interesting question to about if it's spectrum versus, you know, just the time lead.

UNIDENTIFIED FEMALE: Misdiagnosed.

DR. JEFFREY P. BROSCO: Yeah. Yeah.

Undiagnosed because the --

DR. SUSAN BERRY: So this is Sue again. What I would say about that is we don't -- I hear loud and clear, and I understand that early detection could prevent things. And that's a valuable thing, but we simply don’t know about the spectrum. Is that fair? There might be an equivalent mild version that we don't know about that we pick up. I mean, we did this with all other newborn screening disorders. So I wouldn't be surprised if that happened.

DR. JEFFREY P. BROSCO: Yeah. This is Jeff Brosco again. And part of it is because it's such a rare condition, and there's 300 cases in the last 70 years, as far as we know. So, yeah.

DR. MEI BAKER: Can I add on one thing?

DR. SCOTT M. SHONE: Sure.

DR. JOSEPH BOCCHINI: So I have Mei, and then Beth, and then Kellie.

DR. MEI BAKER: I agree with Sue's point.

It's just because right now we have a majority of the clinical information. If I have the spectrum, if I have a milder case, we don't know. It's why we need a pilot study. So clinically you symptomatically -- you identified. Then we can assess the course.

DR. KELLIE B. KELM: But would we truly assess the course if we identified them early, if they got placed on treatment early. So we will alter the course that we see, right? So we would have to identify them and watch them to see what kind of outcome. Otherwise, we won't know.

DR. BETH TARINI: This is Beth Tarini. I would say that in the sort of frame of screening, I think it goes -- you can pretty much place a high bet on it that your prevalence will go -- you will become more -- the prevalence will become more common when you screen because you will find undetected -- yet undetected or asymptomatic disease. And I think, although I don't have the data in front of me, I'm willing

to bet pretty highly that if you compare the nomination packets to the known prevalence after screening many of these conditions will find a rise.

Now, I don't have that data, but it goes -- it goes with the screening principles in general, what we know from screening in adult non-rare disease as well.

DR. KELLIE B. KELM: Kellie Kelm. So it was interesting talking about the benefits of a pilot in terms of course, for example. The other information that I don't know whether or not it's something that's being worked on that can be gathered that's often helpful for us is what are the benefits of early detection but also treatment. And so, obviously, we didn't hear much about that, but that might be useful information when the nominators come back.

DR. JOSEPH BOCCHINI: Okay. Additional questions or comments?

(No audible response)

All right. Hearing none, our goal is to get back to the nominators as soon as possible with our considerations for those things that need clarification as well as a review of the other essential elements that we need to meet before we come back with a full review. So thank you again very much.

So I think with that, we can take a short break. We're going to start back at 3 o'clock.

DR. CATHARINE RILEY: We can start a little early.

Okay. You want us to come back at 3:15? It's okay to take a half an hour break?

DR. CATHARINE RILEY: Yes. And if I -- and if I could, just a reminder -- this is Catharine Riley -- a reminder the cafeteria does close at 3:00. So if anyone is interested in getting refreshments across the way, the cafeteria closes at 3:00. After that, there is a snack room that's available as well.

And I think we'll reconvene a little

earlier so we can --

DR. JOSEPH BOCCHINI: Yeah. I think so. So we have 20 minutes until 3 o'clock, and we'll just come back at 3:00? Okay. Let's come back at 3:00. Thank you all very much.

(Recess taken from 2:40 p.m. - 3:00 p.m.)

DR. JOSEPH BOCCHINI: All right. So welcome back. We're going to start this session with a presentation by Natasha Bonhomme. Natasha is the organizational representative from the Genetic Alliance. She is the Strategic Strategy Officer for Genetic Alliance and overseas maternal and child health initiatives. Natasha has led a range of programs that focus on public engagement in healthcare, including most recently the Newborn Screening Family Program.

Today she will be sharing highlights from the development, implementation, and impact of Baby's First Test, which served as the newborn screening clearinghouse from September 2011 through August of this year.

So, Natasha, welcome.

MS. NATASHA F. BONHOMME: Thank you so much, and thank you for inviting me to speak with you. I know you all, who have the briefing booklets, know that I sent about a million slides. Don't worry. I'm not going to go over all of them, just a select few. But with that, we can dive right in.

Here is our HRSA-funded language, everything that I'll be presenting to you today was supported through HRSA funds.

So even after having spent an extensive amount of time talking about newborn screening education. The question of why still comes up. We still get phrases like this when we go to meetings, which says, "Why do you need to educate about newborn screening? It just happens. Parents don't have to do anything or ask for it." And I think it's important for us to always remember that, that is still very much a top of mind question when we go out, particularly when

we are seeing healthcare providers or people who are just in maternal and child health in general who are thinking of all the hundreds of other things that they have to educate families about. So that we were thinking of this not just within our vacuum of us here who know why this is so important but also our fellow colleagues.

And I think this quote really addresses kind of why it's important. This is from our -- one of our Consumer Task Force members, and she said, "Thankfully we were quickly educated about MCADD by both our pediatrician and some incredible genetic and metabolic specialists at Children's Hospital. He is going to be fine because everyone knows how his body works differently and can factor that knowledge into his care."

And I think what we all want is that every family who goes through the newborn screening process to end up, at the end of the day, feeling this confident, knowing things are

going to be okay, and that there is education for them as families but also for the care team that will be around them.

So for those of you who do not know, our Baby's First Test is a national newborn screening resource center. It housed the Clearinghouse as laid out by the Newborn Screening Saves Lives Act and the reauthorization of that bill up until this past August, and the main goal is to inform and support families and healthcare providers throughout the newborn screening experience.

We've gone through a range of different iterations, and really this has been in response to the behaviors that we've seen of people actually using the site. We have condition-specific information as well as what each state screens for. You'll see through these different iterations on this screen that, that information has become more and more prominent, and that was based off of the fact that throughout the time that BabysFirstTest.org went live about 80

percent of our traffic goes to those two things. It's very clear that is what people are interested in.

And so we've built the site and rearranged it over the years to really meet the needs of people who are coming there. But we have, I want to say, about 79 different conditions. We work with all 50 states and the newborn screening programs to have their information be up to date and are always really looking to see what are people looking for; what are the questions we're getting? Did I go out, or can you -- oh, sorry. What are the questions we are getting and how do we make sure that the information that's on the site is easily accessible.

So I won't go through all of these stats, but this just shows basically where we have been since 2011 when we launched. We launched September 2011 and during Newborns Training Awareness month. I would say the majority of our

visits have actually come within the past three years, and so we've really seen kind of an uptick in the usage of the site.

And one thing that I think is important here to note is the use of -- or users coming to the site via mobile devices. I would say probably four, four and a half years ago that number hit 50 percent and then really just kept going up. And that, again, was one of the reasons why we thought, okay, it's really important to make sure that the information on the site, of course, is accurate but accessible, knowing that most people are coming to the site via mobile device.

A very concrete example of how that was reflected in the way that we presented our work was that you'll notice, if you go to the site, a lot of our pages are very long. You scroll, and scroll, and scroll through. The reason for that is once you put in pages, each page needs to be refreshed, and so we made that

conscious decision to keep the pages long so that there was only one refresh. Again, just one example of really looking at where people are coming from, how they were accessing our site, and then how that led to decisions we made around our work.

And as I go through this -- and obviously I'll be talking about BabysFirstTest.org -- I really hope this is an example of how, when anyone else is thinking about the type of educational work that they're doing, how they can take the lessons learned from this program and really apply it to what you're doing. To me I think that's the most valuable piece of this work.

So we also have an app, and many people thought, well, you're already online. You already have a responsive website. What do you need an app for? So we really built the app based on -- and this is best practices in building apps; it's not that we thought of it

ourselves -- but really looking at who's coming to your information frequently and repeatedly and what are those behaviors. So it's not necessarily about is this app for parents or for healthcare providers, but more so about is this for someone who's behavior is that they're coming to the site multiple times. And that's really how we framed it, and we kept the information on the app kind of specific to that. Again, what people were coming back to time and time again. And what we heard, especially when we were going out and going to nursing conferences in particular is that people really wanted to see what was being screened for in the neighboring state, if they were seeing babies who had actually been born in that other state, so they could compare and contrast and say, okay, do we need to follow up on this or that. So we saw that, that has been helpful for them.

We have our interactive maps, and one thing that is important to note here is our --

we've had great and very involved steering committee members. And one of the pieces when we were launching our maps that our steering committee members really emphasized was you can't just have a map out there. You can't just have data on these topics in terms of what states are screening for what conditions. Right now I'm showing SMA. But you have to put some context into it, and hence, we have that header that shows when SMA was added to the RUSP.

And we realize that, while we may see a map without that context and understand what that means and why every state isn't screening for SMA at this point, that it was important to remember that the audiences for Baby's First Test, particularly based as the clearinghouse, was almost everyone. If you read the legislation, they list about five or six different audiences. But for us our primary target audience were families, expectant families, and new families, knowing that others would be looking at this too.

We have our resource center, which we opened in 2016. And we had always heard, again, it's a really good example of if you build it, will they come? What will actually happen when you put something like this out there? People had often said we need a place that we can have all the resources that people want to exchange. All the times you send an email with an attachment and then -- but you remembered seeing it on that list serve, but where is it. We should have this housed someplace.

So we built a resource center, and what we've actually found is that the people who are using it the most are actually using it to order materials. So many of the materials that I will be showing in, I think, the next slide.

We have, since 2016, we have had a request for and disseminated over 60,000 pieces of educational material. Some of those coming from states. We do co-branding of our materials, and they can be ordered through here, but many of

them through hospital systems as well as healthcare providers. And that really, to us, showed that though -- I should say I think one of the reasons why there is such a high volume there was because up until August, those materials were free of charge. That always helps.

But it was really interesting for us to try to get some data back to say, okay, well, well did you hear about this? How did you know that this material existed and you could either download it -- downloading is an option -- or order it from us to be printed? And we really saw that people just had found it on the site and really said, well, you guys already do it, so now I don't have to do this. And that was generally the message that we got from the healthcare professionals that were ordering this as well as the more community based organizations.

So here are some examples of our materials, and I'll go through these pretty quickly. But one thing that I wanted to note is

that when we were thinking of educational materials, we really were looking for that first line of how do you raise awareness, and some of the data I'll present a little bit later on will touch on that. But the importance of really is it always about having a parent or healthcare professional, all the ins and outs of newborn screening, or is really just about making sure newborn screening actually happened and some of the key components. So a lot of our materials really focus around there. There are three steps to it. We think it's important that people know that it happens in that 24-hour period, and that you don't need to request it at the hospital, which is why a lot of our materials, like this one, really focus in on those types of details as opposed to getting more granular.

One item that came up with one of our consumer taskforce members was that she said, you know, during the -- during pregnancy and during the prenatal time, you get -- you see this image

of all these tests and what's going to happen week to week and trimester by trimester. Why don't we just add on the newborn screening component to that? And so she crafted this, and we worked with her to get kind of the language accurate. And this has become probably our most popular resource. Again, it puts it within the context that people are already thinking about it. They're already thinking about, oh, first trimester looked like this. Second trimester looked like that.

And while from a healthcare perspective we often try to keep these things very different, there are different types of healthcare providers you see during that time, but from a family perspective, this makes sense because it's just the next step. It's now babies here, and now this is what we do in terms of trying to make sure that the baby is healthy.

And this just shows the back, again, some of our core ideas of what we think are really

important for people to know and then a family story.

So in 2015 we started -- well, I should say 2014 we really started to think about, okay, well, we get a lot of requests for information in Spanish. What would it look like to have a Spanish version of the test? Now, I was thinking about this last night when I was going over my slides, and I was like, wow, if we were to do this today, I think it would be a whole lot easier. The technology and the accuracy of translation services are much better today than they were even just three years ago in just the availability of that. But in 2014, 2015, when we were doing this, we went through and we hired a consultant to basically just translate everything altogether.

We launched the Spanish version of Baby's First Test, and because it wasn't just a direct translates, like just put a Google translate on it. We basically rebuilt the site again, and so we basically went from having one site to two

sites, which meant having one project to two projects, even just within -- just to meet this need that we really saw was really pressing.

So just this year we wanted to see, okay, well, who is coming to the site. Based off of work that we had done previously, we know that the traffic and the visits to Baby's First Test, the English version, is almost split evenly between healthcare providers and families, with a very small slice of people who identified themselves as advocates or industry representatives.

So we wanted to see what does that look like on the Spanish site, and we see here that we have a much larger percentage that identify as parents or family members, which I think you can see makes sense.

So all of those materials I showed earlier as well as any of the other ones that we have created as a team are available in Spanish. And I forgot to mention this on the previous

slide, but all of our materials have been vetted through our Community and Consumer Workgroup, so making sure that people who actually would be the end users of these materials have actually had a chance to comment on it, give us really great feedback about what is accessible, what makes sense. And we've also taken in a lot of feedback.

So one of the big ones, that top right card, we used to have baby bottles and pacifiers on them. That was a no-go for any Department of Health that wanted to use our materials. So you'll see that those are no longer there. Again, we always kind of iterating on the work that we have done based off of the feedback that we hear from really many of you here.

Okay. So let's get into some data. Okay. There we go. So it was important for us to really think about how do we evaluate Baby's First Test, not just as evaluation should be part of every program, but for us to say, you know,

it's always difficult to evaluate an educational effort. Let's see what we can learn through this process and be able to share back with the rest of the community.

Today I'm really going to focus on the third listing. I already talked a little bit about Google analytics, and I think I presented to this Committee a couple years ago about the user survey. But the past two years we've focused with our partners at RTI on really evaluating Baby's First Test from a perspective of user knowledge and awareness as well as really evaluating our partnerships.

So let's start with the website evaluation with parents. As with so many things, we started to get the question of, well, what's the impact? This is great. People think Baby's First Test is nice, but what is the actual impact? What happens when someone actually goes to the site?

So we had 777 women and men, who are

either planning a pregnancy, currently pregnant, or their partner was pregnant. And we collected some information from them. We had them either visit one page on Baby's First Test or a control, and the control was a very popular, very well-known pregnancy-oriented website; and the page we picked was around nutrition because we know that is probably one of -- the number one, if not one of the top five, things that people look at when they realize they're pregnant. They want to know what they should be eating, what their nutrition should look like. And I won't go through all of this since everyone can see it, but this is really kind of the modeling for that research that we did -- or I shouldn't say research -- that evaluation that we did.

One thing that we found that was pretty interesting is that even before seeing either the control site or the Baby's First Test site, two-thirds of participants said they had heard the term newborn screening, and that actually falls

in line with some of the other research that's out there when people have been asked, "Do you know what newborn screening is?"

We did take that a step further to say, okay, for those who said they knew what newborn screening was, could they pick out a definition in a multiple choice, and only a third of them could.

And I think that is really important to note when you're thinking about education. One, what does it look like to educate someone around a topic they say, wow, I don't know anything about that; I should learn more, compared to educating someone about a topic that they're like, oh, yeah, no; I know that. Are they going to pick up that brochure or look at that site if they think, oh, that sounds right; I know what that is?

And frankly, we even have healthcare providers who think they know what newborn screening is but have come up to us and say at conferences,

"Oh, yeah, you know, that prenatal testing." Or, "Yeah, Apgar." We get that all the time.

And you know, I think we should cut people some slack. It's called newborn screening. It's pretty broad. It's pretty -- I don't want to say vague. It can be quite encompassing, but I thought that was interesting and something to think about when you are doing your own educational efforts to really think about where is your audience at, not where do you think they're at, but where do they think they are at. That's not right, proper grammar, but you know what I mean.

So after the groups were exposed either to Baby's First Test page, which was just a general page -- we chose one of our more popular pages -- or the control, we gave them a knowledge test, and lucky for us, happy for us, those who had seen Baby's First Test scored higher than those who had not seen it. Again, it wasn't 13

out of 13 correct, but they had a substantially higher knowledge index on that; and it was statically significant.

I'm not going to go through all of this because I want to make sure that there's time for discussion and questions, but we thought it was important to think about what is the self-efficacy that may come out of an experience with Baby's First Test. And we thought that was important because we didn't want -- we wanted to show our test. Is Baby's First Test just a bunch of information that's online, or does it do something to actually engage with families in particular who are looking at it?

Both the control and the Baby's First Test site scored high on all of these measures, so I think that's important. But it was definitely statistically significant when looking at some of these questions such as I feel more confident in my ability to make informed decisions about newborn health. I feel more

confident in my ability to talk to my doctor about issues around newborn health. I feel confident in my ability to find information about newborn screening.

And one thing that this notes is that it shows that there may -- they feel confident in taking the next step, whether that may be to look for more information, to have a discussion with their healthcare provider, and we think that is a success for Baby's First Test. It's not the end all, be all, but if it encourages someone to go and look at something a little bit deeper or to go and speak to their healthcare provider about it, then that is a move in the right direction.

And again, the last three questions are all about foods and healthy eating, and that's because that's what the control was about.

So some of the implications: We really wanted to have as rigorous as possible an evaluation of the site because we wanted to see, was here an increase in knowledge. I think

there's a lot more evaluation that could be done around that, but this was our first step in that direction. We also saw that it did show that this could be an effective tool, and that even exposure to one page. So remember, the people who we just saw, they only saw one page. They didn't tour around the site. They didn't kind of search around. They only saw the one page we showed them that has general information. But that one page exposure did have an increase in their knowledge. It showed a difference in knowledge compared to looking at another site.

Okay. So I'm going to jump into the healthcare professionals. And this is one thing that through the years people have often said, oh, Baby's First Test is for families. It's just for families. And while that is definitely our primary audience, like I said earlier, we know that nearly 50 percent of us -- it's 47 percent of the traffic coming to the site are from healthcare professionals. We know through the

emails that we get from our general contact form as well as well as through our ask the expert module. We get a lot of questions from healthcare providers. So this is why this part of the evaluation was included.

From the 12 that we spoke with, we heard from them that newborn screening was not a top priority to discuss. Newborn screening wasn't a top priority to discuss. They felt comfortable talking about newborn screening generally, but once you started talking about the conditions themselves or even an abnormal result, there is more hesitancy. They weren't quite as comfortable, which actually ties to some of the discussions we were having this morning in the Education and Training Workgroup.

We also know that they were not necessarily seeking out this resource, which, again, is the thought of when you're thinking about reaching out to a group, are they -- you may be reaching out to them, but are they

reaching back out to you? And what are the strategies that you have to put in place when that is the dynamic?

Once they went through the site, most found it to be useful and thought that it was good to educate both themselves and their patients. Oftentimes we have these discussions around education either for -- and it's important to think about who your audience is, of course, but we kind of keep providers over here and families over here. That doesn't mean -- a lot of times it is the same information that is being sought out. So I think you can kind of do a little bit of that double duty, depending on how things are structured.

I won't go through all of these, but it showed that the healthcare providers, their perceptions was that the site was primarily for parents, but that there could be information for them. And you may ask why are we looking at perceptions, but we know that when you have an educational

material, let's say, if someone perceives it's not for them, then they're not going to engage with it. So we really wanted to test, is this something that would catch the eye of a healthcare provider? Would they think to be drawn in or not? Again, it's an important piece when thinking about the different audiences that you're trying to reach out to. Does the person think this is for them? And that can be the case if there's a material that's too complicated. Then families may think, oh, that's not really for me and may pass by it. So again, just one of those lessons learned that wanting to share with the rest of the community.

So about half of the participants said that they reported learning something new, and that they would want to share this information with their patients. And they also wanted to look at it when they had an out-of-range result, to kind of catch up on what they should be saying.

One piece that came up was that they were looking for -- that they thought it would be very helpful if there was a step-by-step approach to what they should do if there is an out-of-range result; that, that would be something that they would like to see added in, again, tying back to some of the discussions that the Education and Training Workgroup is having as well as the new ad hoc group. I think I was talking about that with this slide. I'm trying to make sure we have enough time.

So lastly with the evaluation was our partner evaluation. We've all -- or many of us, I should say, have had those grants that have the section about collaboration. You should collaborate with your federal partners, your non-federal partners, and what does that look like. We felt that we've done a lot of different types of partnerships. Many of them were informal, but we wanted to be able to capture the effort and energy that went into that; and to really answer

the question of and so what? Is that just a nice to have, or was there something really tangible that came out of it? So that's how we framed the questions for that portion of our evaluation.

So we categorized our partners and looked at them, and then we also did in depth web survey of 15 of those partners. So when we did our inventory, we found that we had 59 organizations, and on the next slide I'll talk about what does that mean; what does a partnership mean.

They were split, as you can see here, with many of them being professional organizations, government agencies, government-related programs, as well as web platforms, and that makes sense. That's where a lot of our efforts went at the beginning. It was really to get buy-in for the clearinghouse. I will say, when the clearinghouse first started, there were some who were skeptical, thought why don't people just go to where the information already is; is there really a need to pull that in? So our

partners here really do reflect kind of the places that we were putting our energies.

And then in terms of the web platform and app developers, though we would love to have everyone come to Baby's First Test, we knew that it was actually better to have solid content that we can then give to other groups that already had a track record that already had -- that were already seen as trusted sources of information. And so that is another place where we put our efforts in in terms of reaching out to those parenting and pregnancy websites that people were already going to and helping them update their content around newborn screening.

This shows the different roles and activities that we've had with our different partners. Most of it has been, like I said, around web content contribution and linking to that. Many of them link to Baby's First Test, but the place that I think many people have not necessarily thought of Baby's First Test was

around education and what we mean by there are webinars and trainings and workshops, things that we've done for a range of different communities. Some around nurses.

We've done a range of different workshops with nursing organizations. We've done trainings. We've done trainings for state newborn training advisory groups. We've had states come to us and say the group that's supposed to be thinking about newborn screening clearly needs a crash course in newborn screening; can you provide that? So we've done that work.

The work that's been increasing, I would say, probably in the last 18 months is capacity building and technical assistance. So all of those materials that I showed earlier, customizing them for states and also for different healthcare systems, having people come to us and ask us to review their educational programming and their educational strategies.

And I'll just loop in program evaluation with that. We think that's great. We think that means education is on people's radars. It does not necessarily mean the program originally was built to do that, and so again, I think that maybe just a gap or something to think about in terms of the needs that's out there.

So when we're looking at the value of partnership, what we saw is that many -- almost every partner survey participant emphasized the value of their access to the information. It wasn't necessarily just us, but just the fact that there was a place to go to get quality information that they can pull and form their own educational strategies and saying that in having access to this is what increased their capacity.

Here is just some of the data, looking to see in terms of comparing to other organizations, was this a valuable partnership and what does that look like. These were also all anonymous. So I can't say who said what, but we wanted that

to be -- we wanted people to be as transparent as possible and to see what was the role of babies first test in accomplishing their education goals. And as you can see, everything was typically around strongly agree or agree -- or I should say the majority.

I won't go through these future plans, but we'll emphasize the one piece that definitely came up is that people said they were happy to continue partnering with us, but it really had a lot to do with continued funding. And that makes sense. Like anything else -- we've seen when the newborn screening community has put in both resources from people power to funding, that we've really been able to see a lot of change. And people felt that they would able to continue and maybe build upon this partnership if they actually had the funding to do that.

So in this last section, thinking about lessons learned, through Baby's First Test we have done a lot, a lot of different things, a lot

of things I didn't even touch on today from focus groups, to billboards, to a whole range of different things to think about how do we get out to people; how do we raise awareness. And we've learned a lot in that. I think one of the key pieces that we have learned is you can't just say education and have that be blanket. It's really educating who about what and why. What are the real outcomes we want out of that and to be as specific as possible.

I feel like now I can't do a presentation without having some version of this slide in here. These words are not synonymous. They mean very different things. They are different strategies, and they are different metrics to determine if you are successful or not.

You should not go and implement awareness strategies and think you're going to get outcomes that you would get from an ongoing long-term engagement with an organization or with a community of people. And I think that is one

thing that Baby's First Test has been able to bring to the different partners that we've worked with in terms of asking these questions when we're doing the program evaluation or kind of the capacity building work to say, okay, what is your end goal? Do you just want people to recognize the word "newborn screening" and have some sense of what that means? Or are you actually trying to see a certain change in behavior with, let's say, you know, hospital -- with nurses in the hospital. Okay. Depending on that, we'll have a range of different strategies to fit that.

I won't go into too much detail about ask an expert, but one thing that I think is important is for a site that is very clearly all newborn screening all the time, we still have almost 30 percent of the questions coming in about non-newborn screening related items. And I think that's important for us to think about; well, what does that mean as a program that, yes, we're in the newborn screening world, but we're

in the maternal and child health role. We're in the public health world. What does that look like?

I think it's also telling -- this is only data up to 2017. We have not incorporated our 2018 data in this slide, but the majority of those non-newborn screening questions had to do with drug testing. Will my baby get drug-tested? If my baby is drug-tested, will my baby get taken away? And I think that really reflects where we are when thinking about maternal and child health issues. And so we, obviously, orient people to their state and to their healthcare provider. But it's very clear. I think some people would say, well, if I wanted my healthcare provider to know, I would have asked them that. But again, what does this mean as we think so often just about newborn screening, but again, to a parent it's about what's going to happen with me and my baby and to really be thoughtful about that.

So lastly in terms of lessons learned, we

know that both parents and healthcare providers want to know the importance of newborn screening. While very, very few people actually take two days to sit and think and talk about newborn screening, one of the things that constantly came up when we did focus groups, both at the -- at AWHONN, which is the Association of Women's Health, Obstetric and Neonatal Nurses, as well as focus groups at the American College of Nurse Midwives. At the end they would always say, you know, I've never just sat for an hour and a half and thought about newborn screening and why it's important. I just never -- which makes sense. But then after that they said, wow, like I'm really glad I had this time to sit and think about this and to realize what happens with that filter paper is really important.

We know people want to know about screening procedures, what are the state requirements as well as detailed information about conditions. We know families want a copy

of the results, and they want to know how should they be raising certain issues or questions with their health professional.

And we know that the healthcare professionals want communication tools. What's the messaging? How do I say this? And, yes, even in the interviews that we did with healthcare professionals, there were a few who said, "I don't think this site is very useful for me. I know what I need to say." Great. But then there are also other people who said, "It would be really great to know what I'm supposed to communicate out."

So lastly, I would like to acknowledge the amazing team behind Baby's First Test. I'm happy that I got to present to you, but it has been quite a team effort and really everyone in this room. Every time someone has asked a question here, we've gone back and said is there something relevant to the clearinghouse that we should be looking at. And we've also gone back

and said, well, what can we do to really address the questions that arise in meetings like this, at symposiums, and in different places. So it really was a community effort. I really hope that people feel that way.

And last but not least, it really is about families like this one who go into a situation thinking they don't know what they're going to do, and then at the end of the day they can feel grateful and feel confident that their child is going to get the care that they need. Thanks.

DR. JOSEPH BOCCHINI: Natasha, thank you. That was a really nice overview of all the accomplishments that have occurred during -- from 2011 until now. So thank you.

This is now open for questions/comments.

Scott?

DR. SCOTT M. SHONE: Scott Shone. As loud as I like to be.

So, Tasha, great presentation. Thank you

so much for summarizing Baby's First Test. I have a bunch of questions in which I'll just talk to you about later.

But two -- the last thing that came to mind is we -- and I don't mean to steal Kellie's thunder for her presentation for the lab subgroup tomorrow where we talk about specificity. And so one of the things we talked about on the lab side of all this fun is a lot of the issues we talk about is specificity. What is our target? So in the lab, what's our target? What are we screening for? And it seemed towards the end of your presentation you talked about, well, what are we trying to educate. And you had a slide of what do they want to know. And a lot of the discussions we have here about topics that weren't always on that list.

So based on your experience over the last few years, how -- is it possible and how do you weave in these messages and these topics that, I guess, as a community or as a system we think

that these groups need to know, or maybe they don't and we're -- you know, because part of me thinks that they don't -- it wasn't on the list because maybe they're not aware of it yet, right. And so, you know, thinking about -- again, Mei is going to talk tomorrow about within range results or whatever we're calling it now. How do you think about that?

MS. NATASHA F. BONHOMME: Yeah. That's a great question. You know, I think we have to really think about where people are, and it's one reason why I think in the past kind of couple of years I thought, you know, so many of our discussions around newborn screening are within a newborn screening bubble. And I think that's not a bad thing. I think it really shows, you know, it's a group of people who are focused in on that.

But then when we're really thinking about our audiences that we're trying to reach, newborn screening is one of many, many, many things that

they're looking at. It's kind of like how in so many different educational studies when a group has done an hour long focus group talking about newborn screening, and then at the end they say, oh, is newborn screening important. They're like, oh, yeah, it's really important.

But we've also never gone the next step to say, okay, now in a category of breastfeeding, sleep schedules, and things like that, you know, where is the order. And I shouldn't say we've never done that. I just may not be aware of that information. But again, I think that is something we have to look at.

In terms of the specifics of is it not on their radar because they don't know about it? I think that -- and it goes a little bit to the quote that I had at the beginning, this idea that newborn screening happens. It's just done. Why worry about it? I think if you peel behind that, part of it is aren't there a whole group of other people worrying about that, like once it gets to

newborn screening, once it's prime time, isn't it all set? And I think question mark, maybe, sometimes. And so I do think that that is something for us to think about.

One question I constantly would ask, and then I feel like would get asked back to me is: what is the number one thing families need to know? I think that if as a community we don't come together and have a specific answer to that, then we will kind of always have a range of different educational efforts, and they'll be kind of successful here and probably doing some good stuff here. But really the pulling it all together to say what should be, whether it's a national initiative or what have you around newborn screening education you have to ask that question: What is the top line thing that we need? And it can't be a sentence that's a paragraph long with lots of commas. It really has to be as it's just newborn screening happens. It's here for your baby's health. Is it about --

is it now that it's a -- what was the term from this morning -- a risk assessment. That's different language than just saying, oh, newborn screenings saves lives. It's not that it's counter or opposite to that. It's just a different approach. So I don't think I really fully answered your question, but I think it really is the fact that we really have to think about where these audiences are at and not just where we may or may not wish they could understand.

DR. BETH TARINI: This is Beth Tarini. Thanks, Natasha. That was very helpful, I think, to see the robust work you put together and the thoughtful approach to tailoring it, presenting it, evaluating it, how it can be better, how it can be -- meet essentially the needs of the users.

My question -- or actually my comment is to this issue of specificity -- this issue that Scott brought up, specificity being one issue.

And two points to consider. One - I think that the answer is somewhere in between. I think that the parents -- let's use parents as an example -- on one level sort of do know. I wouldn't say they understand, but the concept of specificity does come up. It just doesn't come up as specificity. It comes up as the fact that this is -- the highest, highest level, 100,000 feet of, you know, the test -- the screening test is not the end-all, be-all. It can be wrong. We need to do confirmatory testing. Like on some basic level, I feel like that is -- that it is a sensitive but not specific test.

And so while we don't get into the numbers and how is it, at the highest level it comes up in a way in which the parents can and perhaps need to understand. That gives them a sense of what the information means and what their actions then have to do to respond to it.

The other point is something that I learned in my decade now being and attending in

the nursery, which is there is this need and compulsion and it's real, to when you have the person in front of you, want to tell them everything that there is to be about the issue so that they -- that you and they feel informed and both as deep as you need to go but as wide. And an example I use is -- and this happens, both in the nursery and the first visit. And the parents are like -- can barely see you; can perhaps remember 1 of 10 words you've said to them. And I've watched the trainees go through this list of like, okay, so when you go home, here's the things I want you to know. And I sit in the rocking chair because it's so long, and I just rock and wait for them to finish. And it's like 10 minutes, and the parents are like "uh-huh" and nodding. They don't remember but one of it.

And it's this whole concept of meeting them where they are. We can go over broad spectrum, broad topics on a list, and then I come back and focus on the one that's the most

important to come back on when they come to clinic. I don't talk on the first visit about everything, just what I just want them to know, and I keep going over it. So it builds -- now I have the luxury of continuity because I'm primary care to be build a bit of a seam, if you will. But I would argue, we do have a system of handoffs, so in some ways we can build that, if done correctly in the system. But I can't trust, of course, that the next person is going to do it because I know I'm going to, of course, always do it when they come back to me.

But there is this sense of when we overload them to some degree, we actually -- we lose more than I think we gain. And I wouldn't say we do that always, but that tendency I don't think bears out as much success as we would like to be. So I think in some cases, less is more. The question is what is that less and how do you frame it. But that I think is an important point.

MS. NATASHA F. BONHOMME: Yeah. I mean, I would add to that, that even with your category of parents, that looks really different depending on what you mean by "parents." Do you mean someone who is expecting and in the third trimester and really is trying to figure out car seat, and clothes, and like what's the plan going to be compared to someone who is like I have the baby; they're healthy; it's fine; can I go home? To someone who is like, why did I just get a phone call about a condition I can't even spell; what are my next steps? Those are all really different.

And I think within that I think you're absolutely right, you know, hoping that there's that continuity of not just care but of communication. But then being able to point people to what's called, you know, just in time information. And a lot of what we've built through Baby's First Test is that just in time.

Yes. We would love everyone to read

this, you know, the second they're like, oh, I'm pregnant. Let me go learn about newborn screening. But then we go back to reality and realize that people are going to look at this when it's just in time for them, and that may be a little bit different per person, you know. Some people are information and knowledge seekers, and will want to know everything up ahead. And then there are other people who are like, what do I need to do tomorrow. Okay. And then when I get to tomorrow, I'll ask you what I need to do after that.

DR. JOSEPH BOCCHINI: So Cindy is next, but before she talks, could I ask the operator to open the lines for the org reps, the organizational representatives on the call. And then we're going to open this up to the organizational representatives as well.

Cindy?

DR. CYNTHIA POWELL: Cindy Powell.

MS. NATASHA F. BONHOMME: Wow, so

powerful. I'm now worried about this question.

DR. CYNTHIA POWELL: It's a great resource that I utilized on many occasions. I would imagine that some of the questions you get might be better addressed ideally, in an ideal world, at the local level. So I wonder, do you have contacts, you know, among the state newborn screening programs --

MS. NATASHA F. BONHOMME: Yes.

DR. CYNTHIA POWELL: -- that you can talk to, not only to help the family but also maybe as feedback to where they might be able to do a better job?

MS. NATASHA F. BONHOMME: Yes. We -- for many of the questions that come in, we usually respond back and say, you know, what state are you in, and have you reached out to your healthcare provider. And then based off of the answers to that, we will either connect someone with their -- with their state. We will give the state program -- usually it's a follow-up

program, but -- or just usually whoever I talk to the most there and say, hey, we have this question. How do you want us to handle it?

And depending on the question, sometimes the state will just give me a pretty direct answer, and then I can communicate that back to the person who sent in the question, I don't want to say family because, like I said, we do get questions from healthcare providers. Sometimes it's, oh, just make the connection between the two of us, and we're happy to do that.

But, yes, I think it does show that there are these connections that may be there on paper and maybe for many people, but not for every single person.

And then there are certain questions -- this would be an interesting way to analyze the questions that have come in, that it's clear -- the answers are out there. They're either on the site or what have you. They really just want a person either to verify or to kind of have that

more -- I don't know -- not necessary one-on-one connection, but feel like they're asking a person a question. So there are a range of different approaches to that.

But, yes, when a question should be handled by either someone at the state level or a healthcare provider, we ask the questions to try to direct the person to that, again, because we also don't want to give misinformation. So it's really more so about guiding the person to the information that they need, but many states have had many, many an email from me and phone calls about, hey, I have this question. What do you want me to do with it?

DR. KELLIE B. KELM: Kellie Kelm. I admit I also have looked at the site and find it very informative.

It's interesting the way that you describe and think about people analyzing it, and I wonder whether or not you look at the data versus someone who looks at it once versus people

that might be -- people who return a lot. Because I know we've heard from other products that we regulate that sometimes people really only can sit and digest, you know, up to maybe 20 minutes at a time. They may come back and do more, and I think also about sometimes when I've gotten, you know, complex medical visits, and I get those people who joke about those discharge papers. But sometimes I need them because I can only -- I can only get past what I need to do on the first day, and then I'll come; and I'll look at the other days and sort of tackle that when I get there.

MS. NATASHA F. BONHOMME: Yeah.

DR. KELLIE B. KELM: But I'm wondering how often -- you know, and obviously, maybe your target mainly is people that look at it once and get a snapshot, but you may also have people that are looking for a lot more over time.

MS. NATASHA F. BONHOMME: So we've seen pretty consistently -- I would say for the past

three years -- that 80 percent of the visits to the site are one-time visits. And so we've built things based off of that, based off of that behavior that we've seen. But we do have that 20 percent that's not -- it's not nothing. That's something definitely there.

I think we could probably -- it would be a really interesting project to look and try to decipher, okay, what are those two different groups? What's their makeup? What does that look like? That was one thing that we definitely learned from working with our web vendor in terms of trying not to -- they tried to pull us away from thinking about, oh, it's a parent, or it's a healthcare provider but really looking at this is someone who needs information on multiple occasions for whatever reason, or this is someone who is coming once and then leaving and building things out of that.

But I agree, I think that would be an interesting kind of next phase of really looking

at our data and looking at the traffic and saying, okay, well, what does this mean? What are they looking for, and are there tools or improvements that we could make to better meet them?

DR. SCOTT M. SHONE: Scott Shone. So I have a point, but to that -- to what Kellie just said in your response, you had a stat. I think it was the average during was like 90 seconds, and so I wonder if -- it was more eloquent when you said a minute and a half. It sounds longer than 90 seconds. So --

MS. NATASHA F. BONHOMME: Thanks.

DR. SCOTT M. SHONE: But my point is I wonder if you can tease that apart too, you know, sort of the -- because when I go back, I know what I'm looking for now. So I'm in. I'm out and I'm done. So that will skew the people, you know, the people with repeat views, but that contradicts what you just said as these 80 percent are one-time visits.

MS. NATASHA F. BONHOMME: So one of the things that we saw and we talked to our web developer about that saying does this mean people are coming in and then just leaving or what? And they said, you know, to do a full review of that you'd actually have to watch people actually go through -- go through the site and leave. But short of that, that minute and a half or 90 seconds is actually pretty average.

I think a lot of times when we think about going to a site, we either think when we're doing more in-depth research and looking for something or we're on our on phones playing a game and that lasts more than a minute and a half. But really when people are looking for just general information like what is this newborn screening. You can go to our home page information and get that information and leave. It's right there.

So again, I think that's something that there could be some really interesting fancy

computer science analysis of. But when we started to see that, we did go back to our web vendors to say is this something we should be concerned about and talked with our steering committee and the team at HRSA who has been so supportive of this work. And we kind of said -- we said, no, this makes sense. People are finding what they're looking for. They're clicking once or twice, seeing what they need and then going. And it actually does align with when you look at the fact that 80 percent of the traffic going to the site is either looking for state-specific information or condition-specific information. So they're looking for things that are pretty targeted and would come up on that initial search.

DR. SCOTT M. SHONE: Just one last point. You know, being part of the Committee but also prior to being part of the Committee sort of hearing your group talk and then others about education, I certainly learned a great deal about education

and communication. I think one of the stats you showed really summed it up nicely for me, which is the 66 percent had heard the newborn screening but only 33 percent could actually define it.

And I've heard some people just say -- I'm sure you've heard it way more than me -- just educate. Just do education, and education is not the 66 percent. It's the 33 percent, the people who know. So I translate that to a three-year-old too.

But I think that's something important for us to bear in mind when we're thinking about education. It's just not -- yeah. It might be reward -- not rewarding at all. It might be fulfilling to say, well, 66 percent of people have heard newborn screening, but it doesn't make a damn difference if they think it's something completely different than what we --

MS. NATASHA F. BONHOMME: Right.

DR. SCOTT M. SHONE: -- want them to

know. And so I think it's -- you know, it's essential that we realize that the 33 percent statistic is what we should strive for. I mean, we should strive for it be higher, but I mean, that's the measure.

MS. NATASHA F. BONHOMME: Right. And I think -- and I didn't have the questions in the section around like what is newborn screening, but we actually went pretty specific. So you could say is it good if 66 percent of people know that babies get screened for something because I think if that had been one of the options and we had said that was the right option because it's accurate, then that number would have been much higher. So again, it's about what are we really looking for? What do we want people to understand? Do we want them to know that babies get tests before they leave the hospital, or do we want them to know that, you know, some are metabolic, some are metabolic. Some are genetic. Some are this. Some are that. There may be a

second screen depending on what state you're -- you know, like just what is that granularity. And I don't think one is right or wrong. I think we just always have to be really -- when we're doing education to say what is it that we're really -- what is the real aim here, both with -- whether it's families, the public, as well as with health professionals.

DR. JOSEPH BOCCHINI: Org reps?

DR. JED MILLER: Jed Miller, AMCHP. Curious if you have any thoughts about the scenario where parents decline a newborn screen, in particular if you feel the current content would suffice if applied a certain way or if you might, you know, develop specific content for that kind of scenario and also just in general, if that topic came up at all during the focus groups or other activities.

MS. NATASHA F. BONHOMME: Yeah. I could do a whole other hour-long talk on that in particular. So the way that the information was

framed on Baby's First Test was that we wanted people to have access to all the information that's available. So there is information there about refusal. But we really frame it as this is a public health program. This is really important -- you know, it's a good thing to have.

I will say that we have seen an increase in the number of questions, whether it's through our contact form, but more so through our Ask the Expert, about different types of opting out. So that may be I believe in newborn screening; I think it's really great. But I don't want my state to have the blood spot. Do you know a company that will just do newborn screening for me? That's one type of -- they're opting out of the system -- maybe not newborn screening in their mind, but out of that system.

I think that is a place that we haven't had -- you know, there was a question once of should we have the opt-out forms on each state's profile page. I didn't even bring that up

because I knew the reaction that I would get from this community about that -- like is that really the most important thing that should be there.

But again, that, to me, would be something that would be on a case-by-case basis. You know, a state would need to look and see, are you seeing trends go up in refusal, and then do a particular educational campaign and program around that, that we could help pull the right content for and then really have it be targeted in that way, because even refusal, that looks different. Again, it depends on what you're talking about. Is it someone say, I don't want newborn screening at all? Is it someone saying, I want newborn screening but not storage and use? Or is it, I want newborn screening, but I don't want the state involved; I want a company. Those are all different. But yes, definitely, I would say an emerging issue.

DR. JOSEPH BOCCHINI: Other questions from organizational representatives? On the

phone?

Annamarie.

MS. ANNAMARIE SAARINEN: Hi. Annamarie Saarinen, the Newborn Foundation. Thank you so much for this. It's been really fun to watch, since I was at the very first meeting after you got your funding and started doing some of this work.

MS. NATASHA F. BONHOMME: Yeah.

MS. ANNAMARIE SAARINEN: So two points. One, my daughter had a surgery 13 days ago. And to your point made earlier, I literally couldn't remember like when the suture bandages were supposed to fall off versus when I'm supposed to like peel them off, versus when we're supposed to come back in for a follow-up appointment versus when she's supposed to get Tylenol. I literally -- and that's someone who's kind of -- we've been through the mill, right? And we should know how to do this stuff.

But there is a eyes-glaze-over sort of component

that happens when you're in the clinical setting. And this wasn't a new diagnosis yet, but it was still a new and sort of somewhat overwhelming -- a new procedure for us. So I always think we're going to get better at it, but then I'm constantly reminded of where your head space is as a family.

And I think, you know, as a foundation, we certainly refer a lot of families to Baby's First Test as a great place to get the overview of what newborn screening is. I haven't known whether I thought Baby's First Test is the place to send people if they're, you know, recently diagnosed or they're just new to something that's been picked up through newborn screening.

And I'll tell you, even if you try to direct them to what we think might be the best either foundation or advocacy organization, or even clinical site -- like a center of excellence around a disease -- I find, in talking to families, that they need a little bit of both the clinical

piece and the community piece. And as I was looking through the site, again, a little bit after you finished your slides, I wondered about doing a little bit better job of giving them a platform to engage with their peers, because I know for me, with this new diagnosis for Eve, really, the first place I went was to Facebook. And I found those two or three communities of families that have had same diagnosis, around the same age, and the same course of treatment, so that I could feel less alone, and also just to get like the straight talk, right? You want another mom or another dad to say, well, this is what happened to our daughter, you know, two weeks post-op or what have you.

And so I wonder how Baby's First Test -- if not to take on more scope than is required, but just to like do the kind of handoff that you were talking about.

And then with regard to data on opt-outs, I was in Manila a few weeks ago, and I was

reminded once again how different countries use different tool sets to promote newborn screening -- both education, advocacy, and awareness -- but in the Philippines, they're very -- I don't know what to say, like very -- and I say this with deep respect for Dr. Padilla and all those who have worked on newborn screening in Southeast Asia, because their program has truly impacted all of Southeast Asia.

But I mean, they're very out there about showing children, with the consent of the families, that were late diagnoses and what their path has been compared to those who were diagnosed through newborn screening. And that has been their most impactful tool to both explain what newborn screening is and why it's important.

And I know we don't tend to do that here, show a lot. And there may be 10 really good reasons why we don't. But I just wanted to raise as one of the things like, huh, that's an

interesting lesson learned in terms of, you know, adoption and driving -- because their decline rate among those eligible is even lower than -- we have a pretty low decline rate, and theirs is even lower.

MS. NATASHA F. BONHOMME: Yeah.

MS. ANNAMARIE SAARINEN: But thank you for all your work.

MS. NATASHA F. BONHOMME: Yeah. I think those are both really good points. To the first one, we actually recently revised, refreshed our Family Experiences section, with the idea of being able to build that out a little bit more as we get family stories that come in, to help build that community.

I will say it's really -- I think your point is just right. I think in a lot of discussions, we'll hear people say -- you know, I'll say, you know, "health professionals" in that space, to say, "Oh, well, then they're just going to go online." And I always cringe a

little bit about that because I'm like online is where they found their community. Online is what got them through it, and we really shouldn't be dismissing that.

Also knowing that there can be misinformation online, but there can also be that person, like you said, who's really going to answer the question that you have as a parent, that isn't just the clinical piece, but like what does this look like when we get home? When will get home -- all of those pieces.

On each Condition page -- so all of our Condition pages are also reviewed by an advocacy organization as well as a clinical expert on the condition. So we do link out. I will say that's something that we put a lot of effort into when we were first starting up, but something that we're starting to look at again to see what makes sense in this frame; what does that handoff look like. And also for us, how do we determine which group is the right or groups to hand families off

to. It's something that we discuss a good bit. But I agree. I think there's definitely more that can be done there.

Also, raising awareness around healthcare providers to say, actually, going online, if you're finding the right community, and you know where to plug those families in could be a really big, positive change for that family. So that can be good, so I agree with that.

And then your second point about -- again, that's about what is the strategy? What is our goal? I bet if you asked the people in Manila "What is the goal?" It's that "Every baby gets screened." That is a different goal than every parent feels like they had an informed decision about whether their child gets screened or not. Those are different, and there are different strategies that go towards that. And again, it's about identifying: Which is it?

And since you brought up the Philippines, one of the slides I took out is that, though our

traffic has been increasing, which is great, the percentage of people coming in from outside the US is also increasing. I would say probably two years ago, it was closer to 10 to 15 percent. And now we are getting close to 20 or 30 percent of the traffic coming to the site are from other countries that have, you know, good, robust newborn screening programs. The UK, India, and the Philippines really make that up. So it doesn't all just stay here.

DR. JOSEPH BOCCHINI: So Beth, I'm going to give you the last -- yeah. Okay. All right.

Well, I want to thank you, Natasha. Well, we have to move on to the next segment. So thank you. So thank you for the presentation and great discussion afterwards. And I'm sure we'll hear more about the evolution of this over time.

But that brings us to the next section pretty nicely. We're now going to talk a little bit more about educational activities and newborn screening. Certainly, in the last few minutes,

we've highlighted communication, educational tools that were developed by the Education and Training Workgroup. And these tools are now online, or available on the Committee's website. So that's a great accomplishment to get them in there.

The Committee also identified that it would be helpful to know the landscape of current educational activities related to newborn screening. So to begin addressing this, we put together an excellent panel for you today that's going to highlight some of the work that's being done out in the field. And I believe this panel's going to inform us and inform the Committee's discussion from this topic, in particular the work of the Ad Hoc Committee that we just formed related to potential education issues related to risk communication.

So Dr. Beth Tarini, Committee member and Chair of the Education Training Workgroup, is going to provide an overview of the educational activities

and give us some background for the context of why this is an important topic to be addressing now. And she will introduce our esteemed panel members.

So Beth.

DR. BETH TARINI: Thank you, Dr. Bocchini. Thank you to the Committee, and especially to the individuals who I will introduce to you who will share their activities related to education with us today.

So my job here today is to introduce you to the panel and also moderate a question-and-answer at the end. And the goal for the panel is for us to highlight the critical role of education in newborn screening, if we've not convinced you of it already; feature the achievements in ongoing activities in newborn screening education.

Now, these are only some. I'm not saying -- this, by no means, is a comprehensive list, but these are ones that we are aware of and who

have membership, have been active through, in some way, our Committee or workgroup; and also to foster discussion and spark ideas for future collaborations and projects.

And so some background. So I looked up "educate," because having worked with Natasha on Baby's First Test, I have heard many of these tenets before of what's our goal, and what is the goal of education; what are we trying to do. And it's been helping in teaching me along the way.

And so I will tell you this: It's very hard to find definitions that are actually consistent, which probably is not an accident, given the conversations we have here. And a lot of them focus on sort of just handing -- like the one here in Merriam-Webster -- just handing someone -- like to provide you with information.

And just this morning, in our Education and Training Workgroup, we talked about how simply giving someone information, (a) doesn't mean they're actually going to take it --

actually going to input it, digest it, and/or change their behavior. So I think that what seems like a simple topic in concept, at face value, has additional layers of complexity that I hope that we'll continue to appreciate through this panel and after.

And so here are two examples that I wanted to point out of ways in which we have used education, and at every point in the newborn screening process. So on your right, the handout is from Amy Gaviglio in the Minnesota Department of Health. This came out of an infographic of trying to point out how newborn screening works.

And I would say that -- why I wanted to use this is, one, I've always liked this infographic; and two, it talks about educating every piece of the process, right? It's not just about what's the disorder, or what's the out-of-range test mean.

There are pieces of it that are talking about what's happening along the way. Now, I'm

not saying that -- again, based on my previous comment -- that everyone has to be told this at one point in time -- but to appreciate that education happens at each -- can be done and apply to each step in the process that is newborn screening.

And the handout on the left is from Baby's First Test. I didn't pick this; it was Google Images. So I didn't go to Baby's First Test first. It just came as one of the nice options that I could have. And this talks about different -- this is an infographic that I think also points out the issue of communities, bridging people of different language and culture. This is -- if you can't see it in the back -- an infographic about newborn screening in general that is done in Spanish.

And this I wanted to point out -- now, I didn't have to say, I didn't anticipate finding this in the first paragraph when I reread this -- this blueprint for the future that we talked about this morning and also in our workgroup.

And in this call for a national agenda on newborn screening programs -- I believe this is from the early 2000s, from the AAP -- the American Academy of Pediatrics -- they talk about laying out sort of newborn screening moving forward and what they feel are the issues to be addressed.

And in the first paragraph, you can see that they -- in the red box, they say -- this is the second sentence of the entire paper: "This screening takes place within the context of a newborn screening system. It involves the following components: Screening, short-term follow-up, diagnosis, treatment and management, and evaluation." And the last sentence of the paragraph says, in yellow: "Inherent to each of these components is an education process."

So, again, to reinforce that education is part of every step of this process. We tend to often focus on the communication and the result. That is fine, but just to have an appreciation of the education that goes on at all layers of the

process to a variety of stakeholders.

And this is to highlight that this Committee has addressed formerly, in the past January 10 -- Dr. Kemper and colleagues -- members of this Committee at the time, Dr. Howell, the chair -- setting forth a blueprint for maternal and child health primary care education, Gen-X and genomics and the recommendations that came out of this Committee.

So although many of our recommendations, the vast majority, are based on disorders, we have a history of addressing issues related to education, especially as it relates to healthcare providers.

And so a little bit, then, about the workgroup that represents -- or tries to focus and identify issues for the Committee-at-Large. We are one of the three Committee workgroups. We have a history that is, in fact, I think -- I wanted to point this out -- instrumental in moving federal funds.

My understand from Dr. Trotter was that

this workgroup was instrumental in garnering the interest and support for the funding of the Gen-X and Primary Care Institute, which was an award made through HRSA, and the mission of which was to increase primary care provider knowledge and skills and providing genetic-based services -- so that what the workgroup does has ramifications not only for deliverables as it relates to the providers or the stakeholders, but also as it relates to generating new knowledge and resources for the greater population that we serve.

And also to highlight that we are a diverse membership. We span a number of organizations, a number of disciplines, and a myriad of stakeholders. And I think this is our greatest strength. We had this this morning, and I have to say, ironically, one of our best meetings ever, because when someone brings up an issue related to a project or a challenge they're facing in education, we invariably have a stakeholder in the room that can speak to how

their community may or may not have a need, or they may or may not have a value to provide and connect them with someone who has these resources already, or to outreach to their organization. So the identity and expertise and the connections of our membership are quite rich and an exceedingly valuable resource.

So our recent projects -- which I'm sure you remember because you hear about them -- well, maybe not because we continue to inch slowly towards the end. This is one of our two products that is some time in coming, but all good things take time. And so this is the communication guide that was spearheaded by Amy Gaviglio and others, which is now live -- right? -- it's live on the HRSA website.

And this guide is meant to help provide a starting point and a framework -- it's by no means comprehensive -- for providers as they speak with parents about an out-of-range newborn screening result. It is based upon research done

by a genetic alliance, Dr. Carol Greene, as well as expert opinion from stakeholders with expertise and experience in the area. And it is meant to complement and enhance the work done by the ACMG and others on the ACT sheets, which are another resource for providers when speaking with parents about out-of-range results.

So this guide is now live, I can say, on the website and available to be used by others. And hopefully, we can talk about if there are other ways in which they might say -- useful. And it will be brought to now the ACMG and the ACT Committee to see if there are ways in which they think elements of this might be helpful with the ACT sheets.

And this, also known as the matrix -- not the movie, but the tool. What else have we called it? It's now called the Newborn Screening Education Planning Guide, for those who are wondering. This document is a document that's meant to be used as a resource to individuals who

are creating educational materials related to newborn screening. It is meant to be broad and comprehensive in its scope of stakeholders as well as topics.

And it is a guide to say -- based on expert opinion and input from the stakeholders themselves, groups of the stakeholders. On the column on your left -- yes, on the column on your left, you'll have the list of stakeholders that are here, that we've listed -- include parents, prenatal educators, doulas, primary care physicians, condition-specific advocates.

And then, going across the columns, you will have different domains, if you will, of information, such as the benefits of screening; where will the newborn screening results reside permanently; how and when will they be received. And these domains and whether or not they may be germane or of immediate, intense interest to the stakeholders themselves. I should say that this plan and guide -- which is not live, but is soon

to be -- was spearheaded by Cate Walsh Vockley as well as Jeremy Penn. And they've done a tremendous job in getting this to where it is now.

And this guide is meant as a starting point. It is not meant to discuss the content of what would be provided under each of these domains, nor the frame that's most effective in communicating it to the stakeholder, but merely to serve as a foundation and a framework as someone starts to create these educational tools. It would help as a planning guide; hence its name.

So this morning, the Ad Hoc Workgroup met -- that Dr. Bocchini referenced and that members of the E & T Workgroup are participating in. And this is a joint effort, and there are members of the Committee, the Education and Training Workgroup as well as the Laboratory Standards and Procedures Workgroup, and this effort chaired by Dr. Baker, in our goal to address opportunities

and challenges related to interpreting newborn screening results, particularly related to communicating the strengths and limitations as well as educating the different audiences that are involved: Providers, parents, and the public. And so Dr. Baker will talk about that tomorrow; we won't talk about that today.

What we will talk about today -- you will hear from Jackie Seisman, who is the Assistant Director of Maternal and Child Health at Genetic Alliance. And she'll provide a summary from the Education and Engagement Summit.

We'll hear from Dr. Debra Freedenberg, who is the Medical Director of Newborn Screening and Genetics at the Texas State Department of Health Services. And she will provide us with a background on the creation of the X-ALD education materials, particularly a retrospective comparison of the process used with the framework that was developed post this summit I mentioned.

Dr. Susan Berry is a Committee member and

Professor and Director of the Division of Genetics and Metabolism at the University of Minnesota -- will discuss development -- the who, what, why, and how of the newborn screening, more than a PKU screen educational tool, and the user of it in the field.

And Kim Piper, who is the Executive Officer for the Center for Congenital and Inherited Disorders in Iowa, will discuss the use of a deliberative community engagement process to inform Iowa's newborn screening processes and education efforts.

And then we'll have a brief period for Q&A afterwards, when the panel will sit here, about 10, 15 minutes.

DR. CATHARINE RILEY: Hi. Catharine Riley. Dr. Tarini, I was going to see if we could invite the panel members to all come up at this time.

DR. BETH TARINI: Okay.

DR. CATHARINE RILEY: Thank you.

MS. JACKIE SEISMAN: My name is Jackie Seisman. I'm the Assistant Director of Maternal and Child Health Programs at Genetic Alliance. Thank you for the Committee for inviting me to speak today. Today I'll be giving a quick overview of the Beyond the Bloodspot Education Engagement Summit, which happened in June 2017. And this is just a quick disclaimer that this program was supported by HRSA and does not necessarily reflect the views or endorsement by HRSA.

Before I get started, I did want to give a brief acknowledgment to the Summit Task Force. They were instrumental in the planning process, and really, the support and guidance they gave us really helped shape the summit. And also a special thank-you to the 2017 cohort of Consumer Task Force members, who served as family leaders throughout the summit.

So just to give you a little bit of background, the Beyond the Bloodspot Summit took

place over two days, in June 2017, held in Washington, DC, and hosted by Baby's First Test. We did receive a -- this was one of the first, if not the first, first national meetings really focused on newborn screening education.

We received a really diverse number of attendees. So we had state representatives, so 30 representatives from 22 states. We had healthcare professionals, families, industry representatives, and other health communication experts, as well as national organizations and programs.

We had over 107 people register; 90 people attended. And at least one state from each of the seven regional genetics networks attended. And the full list of states should be in your briefing book.

And this is just a snapshot of the organizations that attended. As you can see, we had a lot of healthcare associations as well as public health organizations and maternal and

child health organizations, and family-led.

The goals of the summit were to identify and evaluate best practices to improve family and healthcare provider understanding of newborn screening, to identify best practices to increase family and healthcare provider involvement in the newborn screening system, and finally, to evaluate family and healthcare provider involvement in the newborn screening system.

Now, these, of course, were very ambitious goals that we had. And I would say that by the end of this process, we did not achieve all these goals. But what we found at the end of the summit is that we were able to really show the starting place and accurate representation of that starting place for many people in the newborn screening system around education and engagement. And the summit really served as, you know, a place to start having those conversations. And I'll go through that a little bit more.

We covered a range of topics. So from engaging families through priority target populations, educating in a crisis, as well as priority setting in education, just to name a few. And these topics were chosen and selected by our summit task force as well as through our experience as a program.

Now, I won't go through this in-depth because Natasha already touched on it, but one of the major key takeaways and learnings from the summit attendees was really this newborn screening touchpoints, and what is the difference between education engagement, as well as education. And that was like the number one thing that attendees had mentioned as their key learning takeaway.

So as we wanted to give, you know, attendees an option to really focus on what actually is the best practice, we gave them a worksheet of two pages; this is the front sheet. And as they were discussing these educational

practices, educational strategies, to really be able to think about, in each of their workshops, what's the "why," identify the "why"; identify the "what"; identify the "who"; and identify the "How."

Now, this may look familiar to the Association of Maternal and Child Health Programs, and it's because that we worked with them, and they actually attended the summit and gave a brief presentation on their best practices continuum. Since the newborn screening community does not currently have -- or at least an updated, I would say, framework or guide around newborn screening educational best practices, we wanted to give attendees some kind of context of what is a best practice, and when they're talking about these strategies, if it's not a best practice, how best to get there. And this is AMCHP's model -- from a cutting-edge practice to a best-practice model.

I would say the feedback on this was that

this was helpful, but it wasn't necessarily geared to the newborn screening community. And so a lot of these checkmarks, they said they couldn't check. So they wanted something more geared and a more tailored approach for newborn screening education.

We did conduct an evaluation of the summit with RTI International. Overall, it was very positive. And so this is just regarding whether the summit was well organized, and that the objectives allowed opportunity for connections and networking. And this is more of the logistics in terms of positive feedback around the hotel location, meeting materials, etcetera.

These are just a few more reactions from attendees. I will focus on a few, just saying what they appreciated most was the mixture of people -- the families that were very powerful, and it was great to have them alongside the state programs and the clinicians -- as well as helping to see potential partnerships, and of course, the

key fact that newborn screening people have not necessarily been taught to teach.

A few benefits of the summit -- these were the top three from attendees -- was the focus on education. So there's lots of newborn screening conferences, and there's even newborn screening conferences that have sessions on education. But it was completely different to have a complete conference or summit completely dedicated to education.

The ability to connect with diverse audiences. This was a huge one for many that -- you know, having laboratory, having clinicians, having state programs, having families all in the same room really allowed for different perspectives.

And then, of course, networking. That was one of the key pieces from the summit was that a lot of these people would not be able to connect otherwise, because there's others that attended that were not part of the newborn

screening system, but were part of the maternal and child health programs or part of the national organizations, and be able to learn from one another.

So some key learnings and takeaways. And through that evaluation that I mentioned earlier, that was done for both attendees and facilitators. So for attendees, some of the key takeaways was that benefits of educations and its top challenges, effective communication strategies, how to engage with target audiences, disseminating materials, as well as the different players in engagement education and how they share the same goals, that you don't necessarily need to reinvent the wheel, is that there may be another organization outside of newborn screening, part of the public health, that has done something similar that we can adapt for the newborn screening community.

And then, finally, for the facilitators, although they were running most of the workshops

and programs, what they found was they also learned some things -- so involving key target audiences and material development and evaluating those materials. And that family education is important, but it's also important that healthcare professionals are trained to have meaningful conversations with families. And then finally, being able to connect with partners to conserve limited resources, and know where to find those resources.

So in our evaluation, we also asked summit attendees: Having to get those takeaways and learnings, how are you now going to adapt those or contribute to your professional learning or your personal life? And what we learned was that they had more confidence to make education engagement an organizational priority. There also is more potential for connection and collaboration. So the networking that was made at the summit, they really were able to take those back home and see how can we work with

those different partners, particularly at the national level.

And then better engagement of consumers in the development and evaluation of materials, meaning that they learn from the beginning that families particularly, as well as health professionals that may be a target of their materials must be involved from the very beginning.

So the primary theme for the summit was that it really served as a place to bring together diverse groups to connect and share, and that one of the key feedbacks that we received was that Baby's First Test was an important part of that connection for various stakeholders in education. And that some of what the suggestions were from attendees were that to provide more opportunities like the summit, to develop more materials and resources, as well as provide technical assistance around educational activities.

So in conclusion, the summit indicated a tremendous need for resource and strategy sharing, increasing need for a process towards best practice and a more guided approach to education.

The summit did not reach all of its intended goals, but it did showcase a more accurate representation of where people are starting in education. It provided a space for diverse groups to connect and share. And finally, it inspired confidence for communicating those benefits.

There are several outcomes that many people have mentioned kind of throughout the say that came from the summit, the first one being the summit monograph. This is something that will be designed and disseminated by December of this year. But essentially, that gives an overview of what was learned from each of the sessions at the summit as well as including information on educational and health

communications models and frameworks. So this would be useful for anyone outside of newborn screening as well. And then it will conclude with Baby's First Test newborn screening educational best practices framework, which I'll get to in a minute.

And the, finally, we had a couple of outcomes coming from our State Workgroup and our Best Practices Workgroup. So the State Workgroup members met while attending the summit. And through the conversations at the summit -- and this is just acknowledging those members on the State Workgroup And it was also led by Annie Evans, who is the program coordinator for Baby's First Test and so she led many of these initiatives.

But one of the key conversations that was having is that there needs to be plain-language recommendations for reporting lab results, as well as messaging around using the term -- well, not using the term "PKU." And so through that --

and these should be available in your briefing book. They are being reviewed by HRSA and should be available shortly.

But today I'm just going to give a brief overview of at least the "More than a PKU Screen" fact sheet, or tool. There's many components of this fact sheet. But we wanted to make sure that when we're talking about newborn screening that we're not just talking about the blood test. And so this was an important focus from the State Workgroup, making sure that the heel, heart, and hearing was all included on the front pane and evident. Also, the State Workgroup also thought it was important that the states would be able to their state.

Another key component was including a story. So this is from a state newborn screening of why, really, using newborn screening instead of the term "PKU testing" is really important.

And then, finally, some key takeaways. Through this process, we did have the Michigan

Department of Health test this fact sheet. And so these were some of the questions. When they went to a hospital for an educational session and included our PKU fact sheet, they included these three questions. And focusing on the second question: After reading this handout, do you recognize the importance of refraining from calling a PKU screen? And 100 percent identified as "yes," although they did admit that other people in the hospital do use the term "PKU." And through the return of those result, all positive feedback. And I believe Michigan has more events scheduled to test this fact sheet.

And then finally, to the Best Practices Workgroup. I mentioned this earlier in the presentation about attendees really wanting a more guided approach to education around newborn screening. And so this kind of the Best Practices Workgroup came out of that. And I do want to acknowledge Dr. Aaron Goldenberg and Keri LeBlanc for being co-chairs of this workgroup,

and of course, Annie Evans, our program coordinator, who helped lead it as well.

And through this workgroup, the idea was to take the foundational knowledge that we already have of health education and health communications frameworks and adapt it to the newborn screening needs and community. And so this is just showing a very brief picture of the guiding questions that will lead into the framework. And that framework is within your briefing book. And I believe Debbie will be talking about that a little bit further.

And that's it. I just want to acknowledge the Expecting Health staff as well as the Summit Task Force, the Consumer Task Force members, who served as family leaders, and the Health Resources and Service Administration. Thank you.

DR. DEBRA FREEDENBERG: Hi. Thank you for inviting me to participate in this great educational panel. I do have some disclosures.

One is that I'm representing Texas, not the American Academy of Pediatrics, in this presentation. And the other is that we did receive funding from APHL for this implementation grant, which helped us with our educational activities.

So in Texas, we're big, and we've got lots of folks, and we have a lot of geographic diversity, and our educational processes reflect all of this. We take a very broad-based approach to reaching all stakeholders for our educational efforts. And this includes external. We have about 12,000 folks on a newborn screening LISTSERV. We think it's probably more than the submitters, which that was originally meant to serve, but it's gotten much bigger.

We target primary care providers, specialists, families, and parents, professional societies, including Texas Pediatric Society, Texas Hospital Association, Texas Association of Health Plans, Texas Academy of Family Physicians,

Texas Medical Association, and March of Dimes. And that's through generally existing stakeholder meetings and scheduled calls.

We target information for legislatures and policy analysts, although many of them are already on board by the time we plan implementation. And we also target education as far as our State Newborn Screening Advisory Committee, external.

Internally, we also have a lot of folks who have an interest in the education about newborn screening. So we have education for laboratory personnel, for clinical care coordination, which is our follow-up personnel. But we also design education for our administrative folks, which includes division and section chiefs, briefing documents to the commissioner. So we're really trying to reach all levels, both within our program as well as externally.

So I was going to go through about how we develop some of our educational plans for X-ALD.

And really, for us, it starts with funding being identified for implantation and screening, because we're not going to undertake the very labor-intensive and a lot of the efforts unless we know that we'll be implementing within a reasonable time. So for us, really, it starts with knowing that there will be implementation and screening.

In Texas, both the lab and clinical care coordination are housed within the Department of State Health Services on the same campuses, although we are in different sections, with CCC and Community Health Improvement in Laboratory and Infectious Disease Services. And although the majority of the educational responsibility is with clinical care coordination or the follow-up, all planning is done jointly with the laboratory.

So when we thought about X-ALD, we realized this was different. We'd implemented a number of other conditions, and we felt like the differences were going to need a slightly

different approach. So for X-ALD, we knew that a number of the -- first of all, it was X-Linked, which in itself is different. We knew there'd be multiple family members affected, impacted by this. We also knew that we had a significant number of those identified who would be expected to have late-onset conditions.

And also, internally, we knew that this was going to be a condition that would require multiple specialists. So previously, our model had been metabolics, metabolic docs, pulmonary, CF. We knew that this was going to require us to have both metabolics, neurology, and endocrinology all working together to provide the appropriate treatment and follow-up for these children that are identified and who will eventually be adults.

So to start with, we surveyed what was out there. Texas is not an early adaptive state, which sometimes can work to our benefit. So we looked around to see what's out there. And we

did use some information from the New York State Newborn Screening Program, with permission. And we do thank them. But we spent a lot of time looking. We didn't want to reinvent the wheel if it was already there.

So on the first step, what we did was we brought together national experts and the Texas specialists. So we had a meeting that included Texas pediatric neurologists, pediatric endocrinologists, metabolic docs, and our national experts on year one. We educated them on the natural history, therapeutic interventions, and underlying etiology of X-ALD.

We also educated them on the Texas newborn screening system. And some were new to the system and hadn't involved or had any interaction with newborn screening. And we helped discuss design for preliminary algorithms. Laboratory methodology was discussed as well. And that was kind of memorable, because that was the day of the solar eclipse. So lucky I don't

have slides, but I can blackmail a lot of folks with the solar eclipse slides we have.

And then one year later, we reconvened. And again, this was funded by the APHL grant, as was the first meeting. And this time we brought together an all-Texas team -- a Texas biochemical geneticist, a neurologist, the endocrinologist -- and we refined laboratory and follow-up flows, algorithms, protocols. We also brought in a genetic counselor to provide educational sessions, which turned out to be mainly to follow up staff about the challenges that could be expected by families impacted by newborn screens.

We developed a brochure for families about newborn screening and X-ALD, in English and Spanish, which is what you see up. I do have several copies with me if anybody wants them. They're hot off the press; they showed up from the printer yesterday. So pretty good timing on these. And then, for us, our planned implementation date for X-ALD screening statewide

is September 2019, as funding allows.

So, in addition to these activities, we also undertook additional activities. So, for instance, I gave grand rounds at the Texas Neurological Society, Texas Pediatric Society, and I'm still scheduled to give some grand rounds at some of the Texas Medical Schools related to X-ALD.

And to be honest, we were very fortunate in that the leadership of the Texas Neurologic Society and the Texas pediatric neurologist realized newborn screening was coming down the pipe for them, and wanted to come into the newborn screening tent with their eyes open and as educated as possible. And they actually reached out to us before we ever reached out to them, and that's turned out to be a great partnership, with more to come in the future. We also invited a national expert to come in to do our Texas newborn screening grand rounds on X-ALD, and that's now archived.

One of our educators goes out to professional meetings around the state, at an exhibitor of booth at a variety of meetings, and continues to provide one-on-one education. And then we also partnered with Texas Health Steps, which is a free online CME, to include X-ALD education in our newborn screening modules.

So we did it as broadly as we can. We use State funding, grant funding, and we brought in as many folks as we could.

So having done all of this, we then went back and we beta-tested the Educational Best Practices Framework Guide retrospectively. So we'd already done what we were going to do up until that point. And we went back and we looked to see how closely we fit with the guidelines. And we looked at the guiding questions as well.

So for Texas, the "what" and "why," for us, the really first step was the decision to add X-ALD onto our newborn screening panel, which meant that many of the "what" and "why" questions

were answered before formal educational efforts began. But we still had lots of discussion about how to implement this education. And we went wild. Most of it we figured we couldn't do, but we were talking about developing game apps and all sorts of things, to make it more relevant to the reproductive-age population than some of our more traditional educational outreaches.

The second level of the "who," we had many target audiences with different time lines for education, different approaches, and as I said, a large geographic area.

Internal, as I said, it was lab personnel, follow-up personnel, administrative personnel, external with families and patients, primary care provider specialists, professional partners, and policymakers, administrators, and our Advisory Committee.

So one of the things in education when you put out printed matter is that you really want to have it at a sixth-grade level. For us,

we have statewide guidelines. So everything we put out gets reviewed for both plain language and for accessibility to minority and disabled populations. And the reason that this brochure is not up on the web yet is it's still undergoing the formal review. It's passed for the printed, but the online version is getting reviewed right now. And as I said, we had lots of discussion about the modalities to be utilized.

The third level is the "when" and "how." For us, we developed a comprehensive time line, working backwards from the anticipated implementation date time line developed. And the time line was developed simultaneously with the discussion of target audiences. And really, a lot of the education was dependent on available funding, and we needed multiple funding sources for the education.

So for the future, we're fortunate to receive the CDC grant on newborn screening, new condition, and implementation, capacity-building

and quality improvement through data harmonization. And we are planning on developing an X-ALD webinar.

We had some discussion whether we're developing or revising a communication plan -- a comprehensive plan, but we're in the midst of that.

And in addition, there's an X-ALD tutorial for Texas Health Steps that's being developed. Texas Health Steps is actually Medicaid, where they do provide an online education. It's CME, it's free, it's open to anyone. It's not restricted to Texas; it's not restricted to licensure. So it's available. And so that's now being developed, and it's a tutorial because it's a shorter time frame than a full one-hour CME module for information.

So, really, in summary, we've gone broad as we can. We've needed lots of help and lots of folks, and we're fortunate in that we've had buy-in from our partners and our stakeholders. And

as I said, a couple of these around if anybody wants them. Thank you.

DR. SUSAN BERRY: All right. Thank you so much for the opportunity to share some work. I'm Sue Berry, and I'm actually speaking today on behalf of the Midwest Genetics Network. Just a word to the wise: I'm not actually going to talk about the product that you mentioned, Beth. I'm talking about a different one.

But I wanted to highlight a project that our Midwest Network Provider Education Workgroup has undertaken, which is to do education regarding return of newborn screening results. We also have our disclaimers. See, we're being very good about putting those disclaimers on.

I also want to specifically highlight my collaborators in this activity, most notably mentioning Whitney Thompson, who's a medical student, who had a requirement in our medical school to undertake a quality assurance project. And she has gone so far beyond the expectations,

that if you can sign her up as one of your residents, you better do it. Okay.

So what do families want regarding newborn screening results? And I think we've heard a lot about that. Natasha really carefully explicated that. And this is what we also ended up finding. Of course, most of the time, what we've all talked about -- and here I'm even going to worry about the word -- we focus on the positive results. And even that throws you off, because "positive" sounds good. But in newborn screening, the positive results are bad. So we don't have the right words, and we focus primarily on the positive, which is a rare event.

There's a pretty well-established process for dealing with those abnormal results. That's what we focus on trying to convey. But most of the results people get back are not positive. They're normal or are in range or whatever term you now want to use. And what do we do about those?

So the quality assurance process that Whitney initiated was to learn from families about newborn screening results return. And so she, and partnership with the Minnesota Department of Health, undertook a survey of families regarding the receipt of their results.

In part, this was stimulated by a mom, who told us this: She said she didn't remember receiving her child's results. She had no recall of -- knowing that he'd had a newborn screen -- but sort of being told that no news is good news from somebody in the hospital. And despite a newborn screen that was in range or normal, this child had a diagnosable newborn screening disorder. It turns out it's a hard one to diagnose, and he was very severely affected by his condition at presentation, despite his newborn blood spot screen, which had been normal. Okay. So thank you, Leo. This is, in part, for you.

So what we initiated the project to do

was to see if families received and understood their newborn screening results -- the normal ones. We wanted to find out how those negative newborn screening results were handled. And we were doing clinics in Minnesota, because that was our job. And we wanted to know what improvements were needed for efficient and effective communication of those results to providers and to the families.

So we did a survey. And this is a little copy of the survey. It was a paper survey we sent to parents four weeks after birth, via the Department of Health. Patients were selected -- or the families were selected on people that had negative newborn screening results, and they were in a specific clinic, so we could characterize those clinics.

So we actually sent out more than 1600 surveys. We received a relatively low percentage. And it turns out that it's not that uncommon. I thought that seemed kind of low, but

I am assured that that's not as low as I thought it was. It was 15.6 percent. And what we found out was that more half of the families really didn't get the results back, or didn't know if they'd received the results back. And that was, to us, disconcerting. It turns out that it's probably worse than that. I think were better receiving the results in this group than some.

Okay. So with that in mind, we were able to work with the Midwest Genetics Network, and our Provider Education Workgroup. We brought this information to that workgroup, and that workgroup, which is constituted of primary providers, families, and specialists, decided that one of their priorities for education was to give education to providers regarding the return of newborn screening results.

The vehicle we chose to do this was an MOC quality improvement activity. And the planned activity was to assess return of newborn screening results. So we developed a training

activity that included a virtual learning collaborative, which was a series of three modules regarding newborn screening and return of results. And there are three sessions. This is a work-in-progress. We've actually presented two of these three sessions thus far. And so we're in the first round of our MOC4 quality improvement activity.

The first session was: "What is Newborn Screening?" And it seemed at first that we might not need to do that. But as you have all heard, that is not true. We do need to tell providers what newborn screening is, and it's not drug-testing, necessarily.

We needed to have a session about the return of normal screening results, an activity that receives little attention.

And then finally, we felt that a return of borderline or positive results was essential. That's the place where we will be using one of the tools.

So just a reminder what MOC4 is. There are four parts for maintenance of certification in professional activity. The first one is licensure, or professional standing -- that's MOC1.

MOC2 is what you may be -- more familiarly called as continuing medical education -- or lifelong learning is the priority for that.

In most board certification, there's a cognitive expertise, which is primarily explicated with an exam.

And then finally, most have an expectation for improving professional practice, or quality improvement, which is the MOC4 activity.

So you have to have a quality improvement activity to get Part 4 credit. These can be small or large group, collaborative, web-based; there's lots of different ways to do it. And we ended up doing our initial accreditation through the American Board of Pediatrics for a lot of

reasons.

All right. So we offered our MOC4 through the American Board of Pediatrics. This was a snapshot of the very earliest pieces of information. We'd originally, essentially, paid for 50 pediatricians, and we ended up with 82 pediatricians enrolled. So it was way beyond our expectations, and we are absolutely thrilled that that many people were interested. These are primarily from the upper Midwest, because that was the target -- who's in the Midwest genetics work, but not all. They're from about -- I think it was 26 different practices -- some large, some small. We were very impressed with the broad scope of involvement.

So two of the three learning sessions have been presented thus far. And I'm just going to show you this tiny little snapshot from the first seven clinics that entered data. And I will tell you that the larger data set that we're now accumulating from the initial intake from the

clinics is not much more encouraging.

At first, the gray bars on this are the -- they surveyed to see how many of their patients that were coming to clinic over a certain period of time -- that was the initial screening process -- had actually had screening testing done. And most of the kids had been screened, which was very encouraging, at least. Even in this, you can see that there was one that somebody found nobody had screened. So we went, "Ah." Even one kid lost is one kid at risk.

More concerning to us is the percent of newborn screening tests that are communicated to parents. And one clinic was doing magnificently and conveyed 90 percent of their results back. But most of them were doing very few, and many of them, none, which we found shocking. And it has not improved in that initial phase as we've accumulated more clinics. It's something like this. There are a few clinics where it's a practice, but many of them, nobody gives the

result back.

Okay. So we actually, in Minnesota, extended our Quality Assurance project by creating a newborn screening fact sheet. And we went to just a couple clinics. The student couldn't do everything, so she actually went to just two or three clinics that expressed an interesting in trying to improve their practices, and took the data from those clinics initially. And those were -- it's slightly different, the numbers, than the ones I showed you because those this is only the numbers from those two clinics. But you can see that the distribution's not that different from what I showed you initially, which was many of the people whose parents were from that clinic had not received or didn't know about their results.

We gave a brief introduction about using this newborn screening fact sheet that basically has some information about what the screen is, what screens are and what they're not, what it

means to have a normal screen, to be somewhat reassuring, but also to remind people that you should never assume that the screen is the only thing that gets the -- you have to strike that tone; we've heard about that.

Afterwards, we surveyed those same clinics and the babies that were born in those clinics, and found a remarkably increased number of patients who had received or believed that they understood their newborn screening results more effectively. So we were really encouraged by that, as we move forward with MOC4 activity, that it won't take a lot of effort to get a big impact in sharing that information, because we were really pleased, even in this little snapshot pilot survey.

All right. So our Session 3 that we'll be doing is return of positive results. And I'm preaching to the choir when I talk to the folks here about the utility of education in these. An idea is to improve the sense of engagement and

trust from families, to improve quality of care, to respect patient autonomy, and to use medical resources wisely. The ultimate goal is to improve satisfaction of the families and the care that their children receive.

And we plan to frame the return of positive or borderline results around the tool -- the communication guide that's been provided. The module that's being prepared will be focused using this. We're not going to try and teach providers about each of the disorders. We're going to teach them and talk to them about how they share this kind of high-anxiety result -- high anxiety for the provider and for the family. And this tool, we think, will be very effective in conveying that.

So with that in mind, we will complete this initial MOC4 cycle. We can't wait to see how it turns out. We really hope it will be helpful to the providers who've participated in the process, and that it will impact their

practice. We plan to be able to try and certify this MOC4 activity for additional professionals, not just pediatricians. And we want to see how the initial participants improve their practice to the degree that that's accessible to us. The MOC4 tools are pretty powerful in being able to do that.

We're hoping to sort of have an advanced practices in newborn screening information module. That's a little further along in our planning process. And of course, we want to make all of these education materials as broadly available to the community as we can, not necessarily tied directly to performing MOC. So all of the presentations that we're doing for this virtual learning collaborative will be posted on our website for the Midwest Genetics Network, and available for anyone to view that will find that material helpful.

And with that, I will turn the podium over to Kim. Thank you.

MS. KIMBERLY NOBLE PIPER: I'm Kim Piper with the Iowa Department of Public Health, and I'm going to share with you a little bit about something we've been working on for the past couple of years: A Deliberative Community Engagement project. And I'm going to specifically speak to the recommendations we got from the participants in that project -- the recommendations we got from them for communicating newborn screening information and reporting results.

By way of background, when we talk about adding genetic and inherited conditions with late-onset types, potentially higher false positive rates, and treatments with a high cost and potentially devastating side effects, this poses challenges that the Iowa Newborn Screening Program needed to address, as it provides mandatory newborn screening for Iowa's newborns.

Advocates, providers, commercial interests, and families affected by these

conditions have a prominent voice. And these voices are compassionate and compelling about what their recommendations are for screening for these conditions.

So in Iowa, our leadership wanted to hear from Iowans that weren't directly affected by these conditions, didn't have anything invested in newborn screening programming in particular, in order to obtain objective, deliberative recommendations for Iowa's newborn screening processes that best reflect the values of Iowans. So hence, we undertook the Iowa Deliberative Community Engagement for Newborn Screening Project.

So a little bit about what is deliberative community engagement. A definition is that deliberative community engagement is a distinctive approach to involving people and decision-making. And it's different from other forms of engagement, like focus groups or town halls, in that it's about giving participants

time to consider and discuss an issue in-depth before they come to a considered view or to consensus.

It is deliberative, obviously. That's purposeful and thoughtful consideration. And deliberative processes can improve the quality of decision-making and engage the broad community and the policy development process. They can be also used to resolve divisive issues and generate discussion about big-picture policy issues, such as adding conditions to newborn screening panels.

And there's a quote from Dr. Michele Gornick, our lead DCE facilitator, that "DCE is possibly the closest that everyday people can get to actually influencing policy." And it's powerful because policymakers receive educated recommendations from their own constituents. And I have to say that I've been contacted by legislators about conditions that they're considering introducing legislation for. And when we say, "Hold off a little bit. We're

talking to your constituents. We're already reaching out to them. Let's wait until we have some information from what they think." And they're like, "Oh, that's wonderful." So, so far we've been able to work through that process.

Deliberative community engagement in Iowa. We recruited a sample of 30 Iowans from across the state. We ended up having two no-shows. They came together at a location in Central Iowa over a weekend. So we had 28 people participate in a weekend event, where they were charged with deliberating newborn screening and making recommendations specifically about Pompe, MPS 1, and X-ALD.

We asked them to consider three specific questions through their deliberation. Question 1 was: What are important factors to consider when planning for future additions or changes to Iowa's Newborn Screening Panel?

The second question is: How should the Iowa Department of Public Health and the Newborn Screening

Program change communications to families?

And then: How can the department continue engaging the public to provide ongoing feedback for the Newborn Screening Program?

And what we heard -- so I'm going to talk specifically about question 2, and that was: How should the Iowa Department of Public Health and the Newborn Screening Program change communications to families?

So we did receive several recommendations about the timing of education. And as no surprise to everybody here, overwhelmingly, they felt that that information should be provided prenatally. They also gave us recommendations about the medium that we should use to provide that education, different approaches that we could take, and then specifically, the content of that information. And this was about general newborn screening in addition to those three specific conditions.

They also gave us suggestions for provider education and information, such as who should communicate abnormal results or positive results; how to communicate results in general; and that we should report all results, not just the positive results. And that echoes what we've heard from everybody so far today.

As far as the content of communication goes, they wanted us to provide information about early versus late onset, what that is. They wanted us to talk about false positives. They wanted us to talk about opting out. And we heard a lot about that, that -- with these new conditions that are more complex, they felt that families needed to make sure that they had the opportunity to make an informed decision about whether to participate in the Newborn Screening Program or not. So they said we ought to be informed that we can opt out, rather than just finding out, "Oh, by the way, you can opt out," because we don't include that in any of our

educational materials right now.

And they also wanted information about the availability and effectiveness of various treatments and how much things were going to cost.

And a couple of quotes from the participants. One woman said, "We also need to include this in prenatal information."

And "Right now, it's like how they give that packet of information to the new mothers, but then the new mothers are probably stressing about their newborns, and don't have time to read that packet. So I think that should be stressed and have that information be provided while I'm pregnant."

And then another person said, "I think it should be done a couple of times, even if it gets close to delivery. And then ask if they have any questions because people are not educated enough at that level."

So that was something that we did hear was document that you provided the information to

them; document any questions or follow-up conversations that you've had with them as well.

The who, what, where, when, and how of general newborn screening education, some things that we heard: Specifically, the "who" -- who can communicate general newborn screening education. Of course, the Newborn Screening Program staff, prenatal educators, prenatal care providers, local maternal and child health programs, such as the State Title V Public Health Agency WIC programs.

We had a participant who got a new job in a WIC clinic, and she said, "There's no reason why we can't tell our clients about newborn screening as well, and we want to. And then, of course, the hospital staff.

And what should they communicate? Again, early versus late-onset types, what does that mean; false positive; the ability to opt out, at least in Iowa; the availability and effectiveness of treatment; how much things are going to cost;

and specifically that newborn screening is not intended to screen for late-onset types. And their discussions about, where when you're talking about screening for late-onset types, that's not newborn screening. "So you shouldn't screen for late-onset types, right?" Is what they said.

And then, when we speak to "where," in the public setting, through PSAs, flyers, presentation to civil organizations, high schools. We heard that more than once, that you should be talking about this in high schools, even middle schools. And we're like, okay.

And of course, social media -- we would be encouraging that. But they said the general public needs to be made aware of this. And I can say that we now have 30 more newborn screening advocates across Iowa through this process.

Where should this information happen? Again, it goes on to the prenatal provider clinics, childbirth education classes, and the

hospitals, and not just in the maternity ward, but everywhere through the hospitals, and health organizations.

And then they also mentioned an outpatient lab while getting their glucose tolerance testing done. So they said you're sitting there, waiting for, you know, your next lab draw. Why can't you get the information while you're waiting there?

When? Obviously, prenatally. That was the big one. And do it more than once, and document that the information was delivered.

And then how? They were okay with the brochures. They liked the brochures -- not specifically the way they were written out, but the idea of using brochures as the primary means to communicate the information.

They like online. And it should be interactive, with the ability to get questions answered.

And videos were fine too.

Some of them said, "No more apps. We already have too many apps. Nothing else will fit on my phone." What we are trying to do in Iowa, then, something that we have in process that's dependent on resources, is we are enhancing our web page for newborn screening that will have education information materials for parents and families as well as providers.

And it's going to be formatted for viewing on mobile apps. And we decided to do that instead of an app, because apps cost money to update, and you have to have them on more than one platform, etcetera, etcetera. So we thought we would just use our web pages, so people can visit those websites as well as view them on their app.

And then we're going to hand out almost like business-size cards that have a QR code and the IP address for the websites, that the prenatal care providers can give to their clients and say, "Here. Go to this site. Learn about

newborn screening education. I'm going to ask you about it your next visit." So that kind of takes the responsibility off them for knowing everything that they feel they're expected to know about newborn screening. No more apps. That surprised me, especially after hearing from Natasha, when the majority of their visits were on mobile devices. So there you go.

So we're going to move on to reporting results -- the who, what, where, when, and how of reporting results.

So when we asked them about who should communicate abnormal or positive results, they said the PCP. And the reason they said the PCP was because they already have a relationship with their primary care provider, and they trust their primary care provider. But they should that they should have a specialist, or have information from someone knowledgeable about the condition as a backup.

And they specifically don't want to be

passed around to speak with different providers. They don't want to hear from a PCP, then hear from a specialist, then hear from someone calling to schedule appoints and that type. They need to know everything then that they need to know. And it needs to be someone with good communication skills and empathy. And they even suggested requiring providers to attend classes in delivering bad information or bad news, and how to be compassionate and empathetic. So we can all get right on that.

What should be communicated. They said we should know: "What does an abnormal result mean for us?" Is there risk for late onset? And they should know that treatment will or will not be started while we're waiting for confirmatory testing or diagnostic testing. They want to know the resources available to help the family navigate the system or to learn factual information about the condition.

So this goes into a lot of what we said

and heard here today. And they said, "You know we're just going to go to the website -- to Web, the -- right, the internet, and learn this information -- and that may or may not be good. So even if they're going to be directed to the website, make sure you give them a website that has factual information on there. And again, Facebook is another good one. And it would be great if you could know those support groups that are on Facebook or social media that you could refer that patient to, to help them navigate.

Normal results should be reported, for sure, or negative results. And if you're not going to do that, then at least the family should be told that no news is good news, and have that be the case, they said, rather than just not hearing anything about if the testing was even done, or the results of that testing.

And where should the communication take place? They preferred by phone, but definitely in person if you need to collect another specimen

right then or if you need to do other testing. And provide hard copies of information when it's available. And the screen tool is a great one to use for that, and you could also give that to the families, or some kind of information checklist, so they have something they can refer to going forward.

When should the results be reported. This is pretty much understood that it should be as soon as possible, and emergently if it's a time-critical condition.

And then how should the results be reported. They said that you should have a support person present with that family, or try to make sure that they have a support person -- family. Understood that that would take some finesse when you're talking to them about abnormal conditions, and how much they're going to freak out because you said "Make sure you have somebody with you" or kind of like, "Are you sitting down?"

You know, what does that do to people's heart rate? So they said that ideal situation would be with a support person present because you're not going to be remember everything that you were told.

It should be delivered with compassion and empathy. Don't pass them around to different providers or different healthcare providers. Provide hard copies of the information, and use standardized communication guides for checklists. They talked about that a lot, and it was really interesting that Minnesota had been working on a the screen tool ahead of time. And so that was a very nice segue and a dovetail into what we were hearing from our participants. And give a copy to the parent, again.

And that's all I have from our communication part. Thank you for the opportunity to present. And again, our work was supported from a sub-award from APHL through HRSA. Thank you. (Applause)

DR. JOSEPH BOCCHINI: All right.

Well, I want to thank the four of you for excellent presentations. It's started to give us a feel for what's going on in the community.

So as Beth comes back, let's go ahead and open up the phones for the organizational representatives again. And we'll see if there's questions from the Committee or the organizational representatives.

DR. SCOTT M. SHONE: Scott Shone. Excuse me. Yeah. I think this question came on the NewSTEPs LISTSERV at some point. I don't remember what the answers are. And Piper, you might have posed it; I don't know. Or because of your work, you might remember. The question is: How many programs or how many people are engaging with communications scientists when they're developing these messages?

You know, I think about when we're developing laboratory tests, we're talking to chemists, microbiologists, etcetera; follow-up,

genetic counselors, medical geneticists. And then the same people are developing the communication strategies.

And I don't mean like press release people. I mean people who are working in the health communications field, people -- you know, like Baby's First Test has worked with -- is that going on? And if not, how do we maybe encourage that? Or let's have another toolkit. Maybe this group or these -- you know, identify this specialty as, hey, when you're developing these new things, these are the people to talk to while you're also developing your assays and your follow-up algorithms.

MS. KIMBERLY NOBLE PIPER: I'm not aware of any states that are doing anything like that. But that is a good point that you raise. Everything that we put out has to go through our chief information officer. And basically, they just make sure there's not a misplaced comma and things like that.

I did like what Texas is doing, and I was going to ask you, who does your review of your information for readability? I mean, that doesn't get specifically to the content -- oh, this is Kim Piper; I'm sorry -- specifically to the content of the education materials. But --

DR. DEBRA FREEDENBERG: Debbie Freedenberg. So the reviews that are done are done agency-wide, and there's one -- I actually don't know who they are. One of our educators is a web-based educator, and so everything goes off to these folks. And then it comes back either modified, or it's, you know, okay to post or okay to release. So all of it is reviewed by one entity within that agency, within the Department of State Health Services.

Right now it may even be Health and Human Services. To be honest, I'm not certain exactly where it goes. All I know is it goes off, and then it comes back. And the time between going off and coming back may not be close. But I can

check into that for you. I think we probably have a -- the office probably has a specific name. I just don't know what it is.

MS. JACKIE SEISMAN: This is Jackie. I would say from our work-with states, we know from hearing from that education and communications work is the least-funded work, and so that kind of gets left on the wayside. And so a lot of states will come to us asking for that assistance. Of course, that takes time and money for us as well. So that's just something that needs more funding behind it.

DR. BETH TARINI: And then I want to comment on this same topic because, back to my original point about education in each process --

Oh, sorry. Thank you. This is Beth Tarini. Thank you, Sue.

-- that Dr. Gornick, who led the deliberative session, works -- she and her mentor, Dr. Zickmund-Fisher at the University of Michigan, just actually are experts in risk

communication regarding laboratory results. And so they have two projects. One was about redesigning laboratory results as they appear in a portal for EMR so that they can actually be communication more effectively to the providers and the patients.

And in addition, Dr. Gornick has worked through the NHGRI CSER group on designing laboratory reports, so when that they come out of the laboratory to providers, they are user-centered design so that that education piece has framework to it that is informed and designed based on communication principles. So my point is to also just reflect back that there is design of the -- I agree with you in -- communication principles here. But there's communication principles also of the lab reports that come out to the providers and/or to the patients.

MS. JOAN SCOTT: Joan Scott, HRSA. First of all, I congratulate on doing a deliberative democracy approach. It is labor intensive,

having done it in other previous lifetimes. But you are able to unpack a lot of stuff when you've got people engaged over a long period of time -- be it a weekend or over a couple of sessions or whatever.

But what had struck me is that what the messages that we hear, regardless of the methodology that we are using to query people -- whether it's, you know, an intensive deliberative democracy approach or a survey or a focus group -- I feel like I'm ringing --

DR. BETH TARINI: No. You're radiating something.

MS. JOAN SCOTT: Well, now I lost my train of thought. And so we are still hearing the same messages about the what, the when, the how, you know. And so it leads me to the question -- and I don't know if you were able to tease this out of the summit -- and getting back to Scott's comment about communication specialists, about, you know, evidence around

best approaches for informing around -- not maternal health issues, even not necessarily just newborn screening -- but around all, you know, maternal and child health pregnancy-related issues.

And you know, is there evidence around best approaches or best practices? Or does that evidence really need to be generated about which is the best approach for what kind of -- and in what setting, if that made sense.

MS. KIMBERLY NOBLE PIPER: That was one of the pleasant surprises of doing this process was that these were 30 Iowans, across demographics -- I mean, we had males who had less than a high school education level; we had PhDs on the Committee -- so wide representation for Iowa -- I mean, as wide as it gets. And they were provided information about the three conditions and the newborn screening system ahead of time to review. And then, when they came together and deliberate, everything they were

talking about -- almost everything -- was stuff that we have spent years trying to reach some kind of consensus or agreement on.

And I was really pleasantly surprised -- I mean, it was affirming to us that we're on the right track. But I think it does warrant -- I mean, this just gives us suggestions and recommendations, and we're still unpacking; we're still doing analyses. So we don't have all of the recommendations ready for public consumption yet. But I do think it's something that could play a part in developing some kind of best practice.

DR. CYNTHIA POWELL: Cindy Powell. My real question is: How do we compete with Google? You know, Mei related a story this morning, and we had a similar experience with our MPS 1 pilot in North Carolina, and Mei's was with the Pompe disease screening.

And I mean, we developed all these, you know, educational resources, pamphlets. We had

the RTI experts, you know, go through them and give us feedback. And we had online webinars that we advertised through our pediatric society. And we would, you know, follow up the phone call to the provider, if there was a positive screen, with, you know, faxing them all the materials and telling them how to get to the website and all that.

And then, you know, in follow-up with families who had positive screens, when asked, you know, "How did your provider give you this information?" And it was, "They told me to google it." So, I mean, it's so frustrating. And I don't know what we can do, really, to get beyond that.

MS. KIMBERLY NOBLE PIPER: I know that you can pay money to have the search results -- your agency rise to the top of the search results. But you know, it's hard to compete with the organizations that do have the resources to do that. But as far as competing with Google,

I'm sure there are a lot of search engines -- agencies that would like to know how to do that too. You wouldn't be saying, "google it."

DR. SUSAN BERRY: I take that as a bit of a rhetorical question, unfortunately.

MS. KIMBERLY NOBLE PIPER: Yes.

DR. BETH TARINI: Although I have to say I don't, because there are times I'll use Google because -- and when I use -- it's not the Google as much as it is the Google to get to what you're getting to. Like just because you google it doesn't mean you necessarily get that information. In fact, sometimes I will google things and explicitly look -- this happens in clinic around formula mixing -- and I will say, "I want to know what Cincinnati Children's Hospital has" -- because I know they must have something. And so I will look for Children's Hospital formula -- so I will google it because it's not at hand, and I don't have it, or my clinic doesn't have it.

And so I would say, first, it's -- to unpack it a little -- when you say "just google it" because I can't -- I don't know, I don't care, I don't have any information to tell you; I'm desperate as the provider -- that's not good. But when I have said -- when someone's googling with a purpose, with a -- and I'm not saying everyone's doing this -- but Google and searching for these resources is not necessarily a bad thing if it's done in the absence of anything else, or if it's also -- and the appreciation of the information you're getting. But I share your exasperation also.

DR. JOSEPH BOCCHINI: Scott.

DR. SCOTT M. SHONE: All right.

Scott Shone. We might be in this generational thing right now where it's a matter of -- I agree with you, Beth, that I think googling is a wonderful resource to use. But what happens now is at least -- in second grade, my seven-year-old and his -- whatever -- I was

going to say informatics, whatever computer class he's in --

DR. BETH TARINI: Yeah. Yeah. It's not about the --

DR. SCOTT M. SHONE: But yeah. But they teach him -- but they're teaching what is a good search --

DR. BETH TARINI: Yeah.

DR. SCOTT M. SHONE: Like it's part of the education now for a seven-year-old --

DR. BETH TARINI: Even --

DR. SCOTT M. SHONE: -- when you look something up --

DR. BETH TARINI: Yeah.

DR. SCOTT M. SHONE: -- what's a resource that you can use versus not. So, you know --

DR. BETH TARINI: There's a gap. Yeah.

DR. SCOTT M. SHONE: -- maybe the 20-something-year-old now who are having children -- that wasn't part of the curriculum then.

DR. BETH TARINI: Yes.

DR. SCOTT M. SHONE: And so maybe this is something that's an issue now. And the target isn't getting away from Google, but somehow educating people what's -- you know, that not everything on Wikipedia is gospel, or on, you know, Dr. Whatever.

DR. BETH TARINI: But I guess, when they google -- where are the handouts, Cindy? Or they keep --

DR. CYNTHIA POWELL: They were online. We faxed them copies. You know, we --

DR. BETH TARINI: So when you google -- when I google like whatever, MPS 1, this -- it wouldn't --

DR. CYNTHIA POWELL: Oh, I mean, I don't know.

DR. BETH TARINI: Oh, okay.

DR. CYNTHIA POWELL: I don't know. I mean, clearly, like the North Carolina --

DR. BETH TARINI: Yeah.

DR. CYNTHIA POWELL: -- pilot study

didn't pop up to the front.

DR. BETH TARINI: Pop up. All right. Well, I will say this, also, about the -- you faxed it with the newborn screening result?

DR. CYNTHIA POWELL: Yes.

DR. BETH TARINI: Yeah, yeah. So when I have worked in the primary care clinic, if it's not in my hand when I walk in the door, or I can't click to get it -- I can't take -- because I'm 10 minutes, 10 minutes, 15 minutes, right? If I can't reach it or click it, it's not there. If I don't have it, and it's really important, I will get up and go get it, if I know it exists, if it's a handout.

So there's this thing about accessibility in these -- and the fax machine, no one sees because it's in the back, and like no one -- and so I get it. But I think that things don't come from the fax machines to the providers. And so there's this other whole piece of the mode of communication and moving the -- in the clinic

that doesn't often happen, which is unfortunate, because the valuable piece is sitting on the fax machine.

DR. DEBRA FREEDENBERG: This is Debbie Freedenberg. So some of the EMRs that are in use now do have pop-ups, and they do -- often, they go to -- UpToDate or just for the minute or whatever -- one of those online databases. And that appears to be a resource that is heavily utilized because it pops up in their EMR --

DR. BETH TARINI: Yeah.

DR. DEBRA FREEDENBERG: -- as they're using it. So if that hospital system or clinic system or whatever subscribes to that and it's in their EMR, that is helpful for the provider. It's not going to help the family per se, because they won't have access to that information originally. It'll have to come through a health professional.

DR. BETH TARINI: Sure, sure. But as opposed to a fax, if you can click it and print

it -- it's a process.

DR. DEBRA FREEDENBERG: Right, right, right.

DR. BETH TARINI: And some of it is a availability and process issue.

DR. DEBRA FREEDENBERG: And so I think getting it integrated into the regular flow is something that will be helpful in the future if it happens -- or globally.

DR. JOSEPH BOCCHINI: So we're going over a little bit, but I want to get Natasha, Mei, and then Bob Ostrander questions.

MS. NATASHA F. BONHOMME: Okay. I know Jackie's been trying to chime in, and she's smarter than me on the --

DR. BETH TARINI: You concede.

MS. NATASHA F. BONHOMME: -- not just this, but many topics. So you should go.

DR. BETH TARINI: You -- she's ceding you -- ceding the floor.

MS. JACKIE SEISMAN: I'll start, and then

you can follow my lead. I was going to say, in terms of reframing your question of how can we beat it and how can we use it, similar to what Scott was saying is that there needs to be some type of relearning of how to use it, what's a trusted resource, what's a not-trusted -- Genetic Lines does have a Trust it or Trash It tool that we often direct people in terms of what's actually trustworthy.

But I would also say there's different ways to -- you know, Google has changed in a lot, and it does take some relearning of what worked 10 years ago, what worked 5 years ago, what worked 1 year ago no longer is the case. And so right now, it used to be that the number of people that used to hit your link used to pop up sooner in the search results.

That has now changed with ads. And now people that are paying for it -- and non-profits, I will say, have up to a limit that they can use for Google ads, and those are the people in the

search results that are coming up quicker. But my suggestion would be, really, around partnerships, and go where people are going. It took a long time -- and I'm saying not me -- Natasha and Melissa were building up Baby's First Test, and we put Baby's First Test linking out to other organizations that are going to -- we know that people aren't always going to Baby's First Test, but they are going to Baby Center, for instance.

And so going where people are searching for information. So it's not as easy as just putting a link -- not saying that you just did that; you put a lot of work -- but just putting a link on a website and hoping it gets traction. So it does take a lot of that outreach and partnership would be my suggestion. And then I'll let Natasha follow that.

MS. NATASHA F. BONHOMME: Yeah. And then the only other thing I wanted to add, really, speaking to what Joan was asking about in terms

of best practices, I think that there is still more digging we need to do to see, you know, what has already been laid out there. But one of the key pieces that we know is that for people to get a message to stick in their head, they need to see at least seven times.

And so, really -- and like seeing that very consistent. It's one of the reasons why we really push making sure healthcare providers are using very similar, if not the same, messages as what we're sending to families, so that it's seeing it all the time. And then you don't really realize when you learned it; you just all of a sudden realize you know it. But I think there's a lot more that we could do in that space.

DR. MEI BAKER: I just want to quick -- adding on -- I think, as these things are popping up already, when we say compete with Google, it really is we want to tell them which site to go, so -- because in the information -- but I think

Beth say something make me thinking it is accessibility, because if you have a print copy, say this address, use it, then they have it -- they don't want to do that.

I think maybe in the future -- I was thinking is electronic report, a newborn screening report. You -- and then report it electronically, you have the link. They can just click down that. I think the family maybe appreciate primary care physician, at the time, who know something, even have address -- they can do -- look more, I think, right.

DR. ROBERT OSTRANDER: So I absolutely agree. I think the just-in-time thing for the primary care doctor is going to be better than trying to disseminate education because most primary care doctors are not going to use their education time for something they're going to see once in a million years, when they're challenged every day by the same thing over and over again. But UpToDate is a need -- you know, kind of

become a lot of folks' go-to.

And since Alex Kemper wrote the Newborn Screening section in UpToDate, it's pretty good. And it also has links to -- I just looked it up -- it has links to Baby's First Test and the ACT sheets right there under Resources. So even though we haven't planned to do this, we've done a lot of this. So there is a good -- if docs go to UpToDate, and they go to the Resources section, Baby's First Test will be there, and the ACT sheets will be there -- I mean, these particular documents are. I also -- just for kicks while I waiting to talk -- looked up X-ALD on UpToDate, and it's not bad.

So if docs go there, if they take the extra time to do that -- so I think, really, one of the big things is, more and more, to make sure that these reliable things are pop-ups, or in the little window, like it is on my computer, where you can get there, because in five -- you know, the time I was waiting to talk between the time

he identified me, yeah, I looked up ELSI and the X-ALD. And that's how we need to function in a primary care practice.

DR. BETH TARINI: And your point is exactly -- go where they go. The doctors will go to UpToDate. And while was going on, I was like, well, where do I go? I go to UpToDate. And so your point is very well taken.

DR. JOSEPH BOCCHINI: All right. Well, with that, again, I want to thank the panel. I want to thank Beth for leading this, and thank everybody for their comments and their participation in today's meeting. We're going to meet again tomorrow morning, 9:30. Any other information?

(No audible response)

Okay. All right. Thank you all very much. That'll conclude today's meeting.

(Meeting concluded)