Transcript: Afternoon Session – April 19

Good afternoon everybody. I hope everybody had a chance to have lunch. Let's do some housekeeping announcements. Please make sure your computers speakers are turned off. Please hold questions and comments until the end of each presentation and when invited to speak, please state your name each time and speak clearly to ensure proper recording for the committee transcript in minutes. Press star zero if you have any problems with your phone line. Members of the public, please make sure you have your computer speakers turned on. Now we will take role, first of the committee members. Don Bailey? Colleen Boyle?

I am here.

Debi Sarkar?

Here.

Denise Doherty?

Here.

Alan Guttmacher?

Here.

Charles Homer?

Here.

Kellie Kelm?

Present.

Fred Lorey? Michael Lu?

Present.

Stephen McDonough?

Here.

Dieter Matern?

Here.

Catherine Wicklund?

Here.

Andrea Williams?

Here.

The organization representatives, American Academy of Family Physicians? Beth Tarini? Michael Watson? Nancy Rose? Association of Maternal and Child Health Programs, Lisa Bruno? Susan Tanksley? Chris Kus?

Here.

Barry Cohen? Natasha Bonhomme?

Here.

Edwin McCain? Carol Greene?

Here.

Let's go back to the committee members, Don Bailey?

Yes, I am here.

Alexis Thompson? Andrea Williams? Fred Lorey?

I am here.

Thank you.

We will start with the afternoon session. The first presentation is an update on CDC activities. This presentation will be by Dr. Cindy Hinton. Dr. Hinton is a health scientist on the Pediatric Genetics team in the CDC's National Center on Birth Defects and Developmental Disabilities.

Dr. Hinton served as a project officer on a pilot project to expand birth defects surveillance and the long-term follow-up of children born with inborn errors of metabolism, identified through state newborn screening programs. She collaborated with the Newborn Screening Translational Research Network on constructing a data set for the newborn screening clinical registry. She has worked with the American Academy of Pediatrics to develop an online program for practice management of newborn screening conditions and is a member of the Secretary's subcommittee on Follow-up and Treatment. Cindy, we welcome you and you are ready to go.

Thank you Dr. Bocchini. I'm really glad to be here to tell you about the many activities going on in our center pertaining to critical congenital heart disease (CCHD). When the secretary endorsed the recommendation for CCHD screening, she assigned tasks to federal agencies, and the CDC was assigned three tasks. The first was to evaluate state surveillance and tracking to monitor the effectiveness of CCHD newborn screening, the second to conduct cost-effective analysis of newborn screening for the early identification of CCHD, and the third to leverage an electronic health record framework for congenital heart defects including CCHD. I want to give you a little update on where we are. I have grouped under those three main categories. First I want to talk about surveillance, public health practice and applied research when looking at CCHD. CDC supports CCHD surveillance and research, and I'm going to tell you about some of the ways we do that, first how we assess state of readiness for CCHD newborn screening, how we support birth defect surveillance programs in states and how we support public health research. Just to remind you, a lot of what I talk about is surveillance data; that is, the routine regular data collection using very systematic processes. Here you can analyze that looking for trends and patterns, and those trends and patterns will often times show something that may be popping out of you, something you want to look at further by conducting epidemiologic research or perhaps prompting some more in-depth clinical research. The information is used to develop policy statements and develop interventions so you can go back to the community or primary care providers, and the system just keeps going for you are doing routine surveillance, looking for patterns and trends and moving on that way.

The first thing we did back when the committee first recommended this to the Secretary was to assess the potential role of state birth defect surveillance programs for screening with CCHD. In 2010, the national birth defects or venture network sent out a survey to all the states, recent to November 2011, after the CCHD was added to the Recommended Uniform Screening Panel. The states were asked about what they saw their roles were and problems they thought they would see. This was published last year, I believe last fall, in the CDC Mortality and Morbidity Weekly Report and newborn screening for congenital heart disease.

There is supposed to be a graph there. Like the barriers identified were things such as the appropriate legislation, funding, lack of an agreement with the newborn screening program, things that have come up as we have heard earlier this morning with the presentation by Dr. Kus. The involvement of state -- has the potential to be implemented by these limited relationships between state defects in newborn screening programs and inadequate staffing and insufficient funds. The recommended can [Indiscernible] -- prior to adoption of screening for CCHD.

This role of evaluation is what programs are looking to where they feel they have the most strength to contribute to a state CCHD newborn screening program. In December 2012, an editorial was published in looking at the types of questions birth defects surveillance programs could be useful in answering. Health outcomes after newborn screening among affected children. Missing primary targets of screening, such as children who were not screened or have false negatives; assessing the burden and screening accuracy for secondary target; the role of altitude, sociodemographic characteristics and other special circumstances; and the contribution of prenatal and clinical diagnosis before newborn screening.

There are specific challenges, the doctor said, for birth defect surveillance programs and want to make sure you have access to the best data sources, that the data are of good quality, that you develop a timeliness of data collection. Birth defect surveillance is not under the same time constraint as newborn screening, [Background Noise]. There was a call for them to work on timeliness of quality data collection. Looking at the aspects of long-term follow-up for comprehensive outcomes, standardizing reporting and working on the aspects of state and national program coordination.

EC has assisted states directly through an avenue we have called -- where in EIS officer will go out and assist the state with an investigation. New Jersey asked when they first initiated their newborn screening. That had to do the rapid implementation. CDC helped them conduct an assessment of how the data would flow, have each hospital could track these children, the electronic health record capabilities at each facility and how could that information be transmitted back to the New Jersey birth defects registry. They worked with the State of New Jersey to pilot a questionnaire for the follow-up of the infants that did not pass the screening and described the epidemiology of those cases detected during the first three months of screening. George asked for an investigation. Georgia has not mandated state screening but became aware of the fact many of the hospitals in the state were beginning to do screening on their own and they wanted some help on how to assess the situation. With the help of an EIS officer, they were able to survey the hospitals, look at the screening flow, electronic health record capabilities and how this information would get communicated back to Georgia. They were interested in

knowing which hospitals were actually starting the screening which had plans to start screening in the next year and which did not have any plans to start adding screening and some of the barriers and challenges encountered. CDC has continued to work with the state of Georgia as they have made their way on how they want to implement CCHD screening. From these publications that were just released yesterday in the blood spot, the rapid implementation of statewide mandate for pulse oximetry newborn screening to detect critical congenital heart defects and assessment of current practices and feasibility of routine screening for critical congenital heart defects in Georgia. I believe this went over, not necessarily out of the field investigation, but we continue to work with New Jersey on the manuscript they are writing under peer review. New Jersey and that the opportunity to look at nine months of newborn screening for CCHD and able to go into more depth about the cases and missed cases and we are hoping for a publication of that sometime within the next several months.

CDC supports states birth defects surveillance program and lends assistance to the national network. Currently we fund 14 state programs within the network; we provide technical assistance with developing and enhancing surveillance systems. We collaborate on epidemiologic survey analysis in pooled surveillance data, including prevalence and survival for many birth defects. Would help the states exchange information, with published state-specific surveillance data annually and when something shows up that looks unusual and if you want to know if something goes on, we will help investigate the cluster evaluations.

Here is a map that shows in green the 14 states where we support State Birth Defects Surveillance and Data Utilization. The red dots I will get to in a bit, but those are centers we have funded in a case-control research project. With the network, we can pull together data and every year they publish in the birth defects research part A and annual report on 41 major birth defects. Each year they choose something to focus on, and this December they focused on CCHD, the prevalence of CCHD by state and type of surveillance systems. States have an active system where they can send abstractors in the field to collect data, a passive surveillance system where data comes in more through administrative records and there is a continuum where states can combine aspects of these.

I wanted to give you a snapshot of the type of data that is reported in this report. We have prevalence per 10,000 live births—by state, the type of CCHD. In addition, they've taken each CCHD list by state and its type of case finding, whether it is active, passive and you can start to compare the prevalence estimates between those. It really is a great report and tool. We supported -- to convene a one-day meeting to support interactions between birth defect surveillance in newborn screening programs. If you recall the survey that went out, one of the

theories that went out was the lack of communication between birth defect surveillance in newborn surveillance programs. What are ways we could help states and programs communicate? AMCHP brought together 12 states (9 were HRSA funded and 3 others), newborn screening programs, Title V directors and NEWSteps. During the course of this day, each program was able to share where they were in the process of initiating CCHD screenings. What were some of the challenges? What are the ways they try to get around these challenges? How could you build collaboration? As a result, AMCHP is writing an issue brief for State quarters and we anticipate this will be ready in early summer.

EC also has a great resource, the Metropolitan Atlanta congenital defect program. This has been conducting active population -based birth defect surveillance since 1968. It is a state program but CDC has given authority to collect these data. The MACDP case definition defines residency in metropolitan Atlanta (reduced from five counties to three counties as of last year); infant or fetus or child that has a major structural chromosomal anomaly present at delivery; infant, fetus or child must have completed at least 20 weeks of gestation; and it is a live birth and the birth defect must've been diagnosed before the child's six birthday. CHD and CCHD are ascertained by MACDP. This is a fairly recent publication that, in addition to the other CHD's, you have the prevalence but then you can also use these data to do other projects when you're identifying patterns and trends. These data have been linked with the national death Index. They can be geocoded with poverty data, sociodemographic data. Coming out, I believe, next week is a report on temporal trends in survival among infants with critical congenital heart effects. Using the MACDP data set, you have access to 1 million births during the 1979-2005 to go. During that time period, approximately 7,000 infants were born with a congenital heart defect and nearly 2,000 of those had a CCHD. It was a large cohort and the analysis the colleagues conducted consisted of survival trends by time period, clinical and other maternal demographic characteristics. Because you have links to the national death Index, you have the ability to know when there was a death at any time during that person’s life. I think that will be interesting. We will send something out over the listserv.

Moving to applied research public health epidemiologic research. As I mentioned, our center supported the national birth defects prevention study starting in 1997. This was an ongoing study in 10 US states. I think there has been some reformulating of it. Live births, stillbirths, terminations of pregnancy. Chromosomal abnormalities in single gene disorders works good. CHD classified cases -- must be confirmed by echocardiography, catheterization, surgery or autopsy. There are three cases for every control, so that is about 30,000 birth defect cases with about 10,000 controls. The controls are live births without major birth defects and selected from either the hospital data hospital data or participating center. There are extensive maternal interviews conducted. Using this data set, researchers are looking at the question of what

proportions of CCHD -- they have operationalized as how they're going to estimate the proportion of live born infants in NBDPS with a CCHD whose condition was detected date and look at the clinical and demographic factors.

In a similar fashion but focusing on Florida, a study that has been supported by Florida March of Dimes is looking at the Florida birth defects registry, linking it to hospital discharge data and assessing mortality and hospital resource utilization among infants with timely discharge versus late CCHD detection and looking at factors looking at timely versus late detection. I think the NBDPS study may maybe going into our CDC clearance which could mean it could be going out for peer review at some time hopefully soon.

Moving onto some of the health economic studies our center has been involved with. There are three studies that have come out from the states. One is a New Jersey cost study. At the same time and EIS officer went to assist New Jersey with assessing hospital data reporting and prevention effectiveness -- they did time motion studies of nurses as they performed pulse oximetry screening and assessed how many resources were being used by the hospital. This manuscript is under peer review. Likewise with the Florida study, Florida service utilization and cost only type versus is under peer review. Then we have an overall cost effective analysis for routine CCHD screening which is also out under peer review. Hopefully soon I will be able to share on the listserv these manuscripts are available.

Another interesting project going on, again not looking directly at CCHD, this is a get CHD overall but they're looking at a subset of the CCHD in this is usually the Healthcare Cost and Utilization Project which is maintained by the Agency for Healthcare Research and Quality. This data set collects discharge level administrative billing data from participating hospitals across the United States. You have principle and secondary diagnoses, procedures, hospital charges, hospital length of stay, expected primary and secondary payer. Within this you have the Kids Inpatient Database and the Nationwide Inpatient Sample which are stratified samples and weighted so when you do the analysis you can interpret as you would a national estimate. The research questions they are asking are what is the healthcare resource utilization of pediatric and or adult congenital heart defect hospital discharges at different ages. How do discharges with critical congenital heart defects differ in healthcare utilization from discharges with noncritical congenital heart defects and what factors such as age, procedure type, insurance status [Background Noise] and again they plan on looking at the CCHD's for these as well.

Leveraging electronic health record, the work going on with this has been done in conjunction with across agencies project. Members of our center have been collaborating with the National Library of Medicine and the National Heart and Lung Institute. Matching AMCHP to various coding systems. The birth defect surveillance programs are used to using a particular type of coding system and that is the way we have been doing it for years. Other projects persist in using another type of code. How do we get these to work together? The goal is to facilitate meaningful data exchange between the various stakeholders, and Dr. Alan Zuckerman will be presenting an abstract at the May newborn screening meeting at Atlanta.

What I've shown you, I've been able to scratch the surface. There are so many staff involved—state health departments, research of families. We have a CCHD work group that meets once a month, I have listed here. Richard Olney had sped up and we share what's going on in the work represented is everything people on this slide have been doing. Thank you.

Cindy, thank you very much. That's a remarkable amount of work being done in a number of states. That is very exciting data. Let's open the presentation up for discussion. First, committee members?

This is Alan Guttmacher, I'm going to have to go off the call and Melissa will be representing me for the rest. Thank you very much.

Questions or comments? Organizational representatives?

Hi, this is Natasha. That was a great presentation. A question I had is for the CCHD meeting I think you said was held at AMCHP and had a number of programs such as NEWSteps. What was discussed about educational efforts?

There was nothing discussed about educational efforts. It was targeted on the defect surveillance program, newborn screening programs, what to do to help these programs work together.

Where would you say has been the place, if you know, where those discussions have taken place around educating either people whose children are identified or even the public in general about the screening?

We definitely have a website that EQ were to use your favorite search engine and look at CDC CCHD, we have an educational website. I can't speak to this with a lot of authority, but I know the people in our center who work specifically with the birth defect surveillance programs work very closely with the programs. The goal of the program is to get people hooked up with services and education. These types of analyses I would say are fairly removed from the education aspect. If you get down to what a state program is doing, it is where more of the education would be going. There is also the heart defect collaborative. They may be doing some educational efforts.

Thank you.

This is Chris Kus, a good presentation Cindy. I wonder any future meeting when would be a right time to hear about the CCHD demonstration Roger asked, the HRSA grants just to throw that out there?

The CCHD grantees I think are going to have a year under their belt in June. I think when they are in year two of their grant cycle or maybe even year three, once they have data or lessons learned they can share, we want to give them a little more time.

Good.

At CDC, some of these projects were new projects specifically geared toward CCHD but because of our strengths in surveillance and epidemiology, it was a lot easier to tap into an ongoing data set or ongoing project and turned the focus toward CCHD. I think there was less of the getting off the ground toward that. Probably within the last year the amount of work that is in turn toward CCHD with available data has really been impressive I would say.

Cindy, it is Chris, and a comment about whether the malformation registry programs are involved or how strongly they are involved with the implementation grantees?

I think it depends on the state and a lot of where they were set up in the communication. In some states I think it has been quite strong. They really have been working together and in some cases taking the lead. I think the brief is going to highlight some case studies. It wasn't a stretch to find some good states to highlight. Birth defect surveillance and newborn screening are working together, and how they solve problems.

This is Joe. Does that sort of speak to the issues you, Chris and Red raised earlier about separation of the laboratory of newborn screening from the hearing and now congenital heart disease? Is it really the birth defects group that is more responsible for when there is a separation in those states?

Let me gather my thoughts on that.

I would jump in; it depends on where the birth defects program is located within the state. Chris can maybe speak to that from New York. Some programs are actually in environmental health in the state health department, relative to more active follow-up.

The other part is how they shared data. One thing birth defects surveillance programs have said we don't operate at the same rapidity the blood spot screening does he know how to go after patient level data. We can come back with that and do it in a timely manner to assure newborns have been screened and look at some of these overall patterns and trends. Even if they are not necessarily housed together, it is having those agreements in how you merge data. New York is a good example of that with their birth defects registry, newborn screening, vital records and intervention and pulling those together, even though they may not be housed in the same place.

This is Chris, what we were doing at hearing screening, in the implementation of that although we have used it to look for information about long-term follow-up. This adds another player that is more involved with CCHD that will make the coordination and more people at the table.

Okay, this does help inform what you are doing as well.

Other questions or comments?

This is Coleen, just one other thought for future committees, I know we had this in January; you are either at a sub group level or at committee level. Staying on top of the early implementers of this to get a better sense of what some of the challenges are, perhaps how the committee can come behind in the help I think is important.

I think that is a good point. I think with the CDC and this committee staying on top of that, I think bringing in other states might be expedited by getting best practices and things like that together from the initiators.

This is Cindy; I wanted to add one thought. I took this out of my slides because I didn't have time, a new project that started, I think, just last year is to conduct surveillance for congenital heart defects in adolescents and adults. We were talking about that you identified them as children, how they are doing as they go through the life course. This is a new project setting, working with a select number of states doing a pilot project and setting up how we conduct this type of surveillance for adolescents and adults. It is starting to get a picture of how these kids are going to be doing in 20 or 30 years.

This is Ed McCabe. There is what is referred to as adults with congenital heart disease clinics. That might be a very good source and I think there are some consortia of those clinics as well.

I think they have been a partnership. I think they are tapping into those.

Okay, good.

Okay, if there are no other questions or comments, let's open the lines to the public and give the public a chance to ask any questions related to this topic.

To ask a question, press star one. At this time I show no questions.

Cindy, thank you for your excellent presentation. We appreciate the work CDC is doing.

Dr. Bertini, we have a comment in the webinar from Harry, what about lost to follow-up with CCHD, it is better than hearing loss screenings?

I guess the question is that it is probably too early to answer that.

Probably so, one of the things about CCHD that makes it different from the hearing loss, these are infants who will be critically ill. I think because they are immediately moved into some kind of special to supervision, I believe it minimizes that immediate loss to follow-up. I cannot speak to what happens further down the road once you get past the immediate crisis, it is not my area of expertise but that could be something I could follow-up with the people here who are more involved with the adolescent or adult surveillance, or have had more experience with birth defect surveillance programs and see how they might be able to answer that.

Also, that a study that is under review with pediatrics in New Jersey might provide is a first glance at that.

Okay, thank you. All right, it looks like we may have someone else typing. This is a comment from Ellen Zuckerman, the Society of Thoracic Surgeons, maybe are fighting follow-up on CCHD across state lines and for long-term outcomes. That is another source, in addition to the adult with congenital heart disease clinics that were earlier described.

The next topic, let's go ahead and close the public comment line, the next topic has no slides, I'm just going to give the brief update. The initial intent of setting up this meeting is we were able if the condition review work was to put the data together before the committee was to sunset or going to hiatus that we wanted to see if we could review Pompe to make a decision. The admonition from the committee is they didn't want to short change the review so we didn't have all the data we needed and time to review before making a decision. After going over things with Dr. Kemper and his group, it turns out the original timeline was a better predictor of what needed to be done to complete the process. We were unable to shorten the timeframe to bring the

condition for review today. The condition review is proceeding in such a way that if it is possible for us to have the main meeting as we would like, that we may be able to have the data in time for review by the committee to make a decision in May. As we said earlier, we will learn more over the next couple of weeks about how quickly the discretionary committee can be chartered so we can go forward. The status of the Pompe review is as follows, and Dr. Kemper is unable to be here, he is out of the country. The evidence review, there are three aspects of the process; the evidence review itself has been completed in terms of identifying the articles and providing the review. I think there is additional work with some of the consultants to finalize that review process. In addition, the decision analysis is underway, that is under the direction of Dr. Lisa [Indiscernible] and that is expected to be completed under the next couple of weeks. The third aspect is the APHL survey for the public health impact assessment. And that is under [Indiscernible]. These processes are going on simultaneously, and it is expected they are likely to be completed in time for the committee to do the review in May if we have the opportunity to do so. I think that is all we wanted to say about that. The goal is to try to have at least two weeks ahead of the scheduled meeting for the committee to review the data.

Are there any questions about that brief update?

Sounds straightforward, thank you.

Next, we have public comments scheduled. I think there are two individuals who wish to make oral comments. Before we turn on the phone line for each of the two individuals, please remember you need to have your speakers turned off when you're on the phone and only have your phone on mute unless you were speaking. If you do not have a mute button, you can use star six. Before you speak, please state your name and organization. The first of the two speakers we have is Dean Suhr who is president of the MLD foundation. Let's open his phone line.

Operator, are we able to do so?

One moment. His line is open.

Thank you. Committee members, I wanted to take a brief moment and touch on three things. I did send a written copy for your reference later on. We want to thank you for your ongoing work

on behalf of those with diseases detectable by newborn screening. I know the work is tedious, but the work you do is important and we recognize that. In these times of budgetary uncertainty, I want you to know there are many of us out here working hard for the renewal of the Newborn Screening Saves Lives Act and we are in support of maintaining the continuity not only for this program but the other aspects that act supports. We are helping to carry that torch for you. The second topic, I talked about my interest in having a renewed conversation about changing criteria for committee approval of newborn screening to not require viable therapy as one of those criteria. I recognize the implications are, in terms of philosophy, focused work and the cost of social services, medical and the impact on advocacy groups. Out here many organizations such as the MLD foundation have come to the conclusion or are coming to the conclusion which might be a question is, knowing your child has a potentially able or serious disease in advance of the symptoms, so you can more adequately prepare for the child's future and then understanding the genetics and how that might affect your existing family is something that is valuable and does that offset this trade-off of having a viable therapy in place? I am encouraging people to start thinking about that. I hope that might be 6 or 12 months out that we actually convene some sort of the caucus to talk about this all is perspective, and please feel free to get in touch with me. I would like to ask education committees consider creating, making us aware in the public if you already have such a document, materials that allow us to work with families and organizations by forcing state legislators to implement screens in advance of advisory committee recommendations. I feel this legislative first approach is a train wreck waiting to happen and makes it more difficult to implement advisory committee recommended screens in the future. If you have any materials that area I would like to share them and digest them. If you don't, I'm happy to participate in creating those materials but I think that is an important topic to work with that committee. That is all, thank you for your time.

Thank you for those important comments, we appreciate your input. Next on the list is Amber Salzman. Amber Salzman is president of the stop ALD foundation. Please open Dr. Salzman's phone line.

Thank you. This is Dr. Amber Salzman and I lead the Stop ALD foundation. As a patient advocate, I was pleased to hear this morning that Secretary Sebelius approved the discretionary committee in place while the Newborn Screening Saves Lives act is up for reauthorization, thus enabling this committee to continue serving such a critical role. The purpose of my comment this afternoon is to provide an update on the newborn screening for ALD with the aspiration of moving the review process forward. At the September 2012 committee meeting, ALD newborn screening nomination was reviewed. The committee recognized ALD and I quote “is a medically important disorder that deserves serious consideration in possessing a well-established case as well as readying diagnostic and treatment protocol.” The committee requested more perspective

data from the pilot study prior to moving forward. Once additional data are available we were encouraged to contact the committee to facilitate an expedited review. The committee would determine if the data merits a formal review of the scientific evidence by the external condition review group. With that as a context, I appreciate the opportunity to review the status of the pilot screening 100,000 California newborns. To date, 50,000 samples have been screened and analyzed. The remaining will be completed by the end of September this year. Of the first 50,000 dreams, there were 12 that tested positive. After those 12-point through a second biochemical screen, only six samples tested positive. Of these 6 samples, 3 came back negative from ALD, 2 samples were female and 1 was male. The other 3 did not have sufficient material to test them, so California is being asked to send additional material. In applying to test 8 out of 8 male newborn control samples came up positive. The bottom line is the ALD newborn screening test has an incredibly low false positive rate. Just looking at the first 50,000, you're looking at a false positive rate of .0024. It correctly identified all the positive control samples in a blinded study and mechanisms are in place to do electro screening on the samples that come up positive I biochemical screen. Since we don't want any more families to unnecessarily suffer the devastation ALD can cause when it is diagnosed too late to intervene, we thought given the -- we help you can provide guidance on how to best work with the committee to move forward the review of ALD. As I'm sure the committee is completely aware, in March this year New York order to implement newborn screening for ALD. That legislation requires the lab to have the test validated in their lab in time to start screening in January 2014. I thank you for your time.

[Captioners Transitioning]

We look forward to the data and we would be more than happy to help work with you to bring that data forward to the committee in the format necessary for review of the condition. As you indicated, the intent was to an expedited review for -- looking at the data when it became available. Thank you for the update.

I don't have any other person listed for comments. Is that correct?

Correct. We had two people signed up.

We now have a scheduled 10 minute break. You can stretch now. Move around a little bit. We should all stay plugged in. Unless there are any other questions at this point, we will take a 10

minute break. I have 1 minute after the hour, so we can come back at 10 minutes after the hour and remain on time. Questions or comments? We will restart at 10 min. after the hour. Thank you very much.

[This event is taking a short break and will reconvene at approximately 2:10 Eastern Time. Captioner standing by.]

It is 10 minutes after. I think if we are ready to go, is Meg Comeau available for her presentation?

Your line is open.

I am available.

Thank you. We want the slides for this presentation.

Hold on.

Are you ready, Meg?

Yes.

Meg is currently the Director of the Catalyst Center at the Boston University School of Public Health. She is dedicated to providing the support to states and stakeholder groups on health care coverage and financing policy for children with special health care needs. In addition to providing strategic leadership to a multidisciplinary team, her work has focused on the role of Medicaid in serving children with disabilities, the implications of federal health care reform for children with a broad spectrum of health care needs and the causes and consequences of financial

hardship among families raising children with special health care needs. We appreciate her presentation today.

Thank you very much. I appreciate the invitation to spend time with you this afternoon. I will share some information on the Affordable Care Act (ACA) and the potential application for people with heritable disorders. I want to thank Lisa Vasquez for advancing the slides. I am using an unfamiliar computer. She will make sure that this is moving along smoothly. I need technical assistance on a regular basis. Having an unfamiliar computer makes it a challenge for me. She will help me and I appreciate it.

Thank you for the introduction. I think we will be able to skip the first slide -- now I have a shameless plug -- a quick overview of the Catalyst Center. The most important thing is that we are based at the Boston University of Public Health and we are funded by the Division of Services for Children with Special Health Care Needs to provide support to you in your work in whatever we began around the outcome measure that all children with health care needs have access to adequate health care coverage for the care they require.

Next --

Before we dive into the details, let's spend a minute they can about the intersection between public health and insurance coverage. They have different but not necessarily competing focuses. Public health is focused on improving population health and insurance coverage is focused on reducing individual financial risk. For example, in this particular area, as state and federal funding has become more inadequate to support the components of newborn screening systems—education, screening, diagnosis, follow-up, management and treatment and quality assurance—there has been an increasing reliance in almost all states on fee for service billing. As the public money has become tighter, private money in terms of insurance coverage and payment has become more important to keeping the system eventually viable. Insurance coverage, both publicly and privately funded, is not just a funding mechanism to keep the trains running. Getting individuals on and keeping them on affordable coverage helps increase broader long-term access to care, which leads to opportunities for improved individual health, which is to opportunities for improved public health and population health.

I think that our interest is a field in the ACA is not just because we are interested in making sure that people have coverage they need, but there are public health implications for all of this which are important.

The ACA -- per pieces of legislation. It is known as the ACA. It is a step in the right direction in terms of being able to move individual and population health forward. I think the ACA holds promise as a gateway to expanding on the goals of improving individual health outcomes and also public health and also offers challenges. Less of what is in it and moreover what is not. We will go over both of these in the next 40 minutes. We'll go over many of the opportunities and some of the potential challenges and then I will welcome your questions at the end. Hopefully, I can answer them.

Next.

There are three major areas of focus. The first is insurance reform -- the patients' bill of rights or the consumer protection provision. The next is new and expanded coverages -- pathways to coverage including expansion, maintenance of effort provisions, and exchanges or marketplaces which are paired with an individual mandate that everyone have coverage. And, there are important cost and quality related provisions. The primary focus in the ACA where the majority of time and attention was spent in getting people covered so that the first piece around insurance reforms and new and expanded pathways to coverage -- reducing uninsured that's the primary goal.

There is much less focus on underinsurance, which is what most children with heritable disorders face.

While this is an historic opportunity, it will not do everything for everyone. For example, what is essential for one group with a specific diagnosis is not necessarily essential for all people. The essential health benefits, which we will talk about any moment, only applied to the individual and small group markets in out of the state exchanges. Most children with health care needs currently get their coverage with a large group or self-funded plans -- Medicaid or Children's Health Insurance Plan (CHIP). The need for the safety net -- for underinsured children -- will continue to be critical especially in these times of economic vulnerability for individuals, families, providers, state, and the federal government.

Let's start with the insurance reform -- reform provisions. These are examples that have particular resonance for people with heritable conditions. The first and most important is the prohibition against denying coverage in the private market based on a pre-existing condition. Previously, insurance plans were allowed to say no to people in an effort to reduce adverse [indiscernible]. This was because they had a pre-existing condition and therefore associated costs. That is no longer allowed for children as of 2010. Starting in January 2014, it will no longer be allowed for adults, either. Dependent coverage is now available up to the age of 26 on their parents plan – in effect as of 2010. The last statistic that I saw is that approximately 6,000,000 young people are now on their parents plan. Previously, many of them may have ended up in the uninsured ranks. That is been a provision that we have already seen a dramatic impact to the positive.

There is no rescission of coverage allowed, regardless of the cost of services used in that way, to affect 2010. Previously, the insurance companies could look back and see when they started to get a heads up that the person might have health problems and they were able to take back the coverage and cancel the coverage and in some cases asked them to pay back the money that the insurance plan had spent on covering their services up to that point. That is no longer allowed. Once you are on coverage, you are allowed to stay and they cannot throw you off because of health status.

Next --

Two other important provisions -- related to guaranteed issue and guaranteed renewal. These will go into effect in 2014. If you are eligible for coverage, and insurance company is required to issue it to you and if you continue to be eligible, they will have to renew it. Section 2705 is a prohibition against health status, and it lists genetic information among the factors that cannot be used in considering eligibility or coverage or premiums effective in 2014. Higher premiums are only allowed based on three factors -- geography, age, and tobacco use. No longer will insurance companies be able to charge people with existing conditions or health care needs a higher premium.

Annual and lifetime benefit limits are another set of important provisions. Effective now, there are no more lifetime limits for existing or new plans. We will no longer see a situation in which a baby is just discharged from the NICU already having reached their lifetime limit and becoming

effectively uninsured as a result. There will be no annual benefit of less than $2 million for plans starting after September of 2012. And, starting in January 2014, there will be no annual benefit cap allowed at all. There is a caveat associated—these provisions are related to expenditures, related to cost. Benefits themselves can still be capped. For example, an insurance policy can say that there are only 20 physical therapy visits for mental health sessions available per year. But, there cannot be a dollar amount associated with them.

There will be new and expanded pathways to coverage under ACA which offers exciting opportunities for people who are previously been unable to get onto insurance or had difficulty affording their insurance. The signature among these provisions is the state exchanges or marketplaces. They are scheduled to open in January 2014 in each state and they are scheduled to open for enrollment in October of this year, which is only six months away. You can imagine that there are is quite a Scrabble going on in the states to make sure that these are up and running. For enrollment in October, when they start to cover people in January. There will be a choice of different individual policies and small group plans. One thing that is important to note is that the exchanges are going to offer policies and plans in the individual and small group markets. They are not applicable to people who have large group insurance access. Small groups are defined as those with less than 100 employees.

There is going to be help for consumers in choosing a plan. I don't know if you have ever had the opportunity to look at your benefit statement or your actual insurance policy, but it usually is about the size of a phone book and it can be condensed into eight point font and difficult to get through. Really, understanding what you are purchasing and what will be covered and what the cost sharing requirements will be. There will be help in making a more informed consumer choice in picking a plan. There is going to be a comparison website where people can look and see the differences between the plans and policies they have available to them in terms of cost sharing and premiums and the actual benefits offered. There will be navigators and people ready to help people wade through some of this and figure out what would be the best plan for them. An incredibly important part of this operation is the tax credits and subsidies available to people with income less than 400% of the federal poverty level. The individual mandate that everyone have coverage requires that everybody get coverage, so the credits and subsidies are designed to help with affordability, to help people that are struggling to afford their insurance or could not afford it previously to be able to get into the insurance market.

The exchanges should help reduce uninsured by increasing access to decent and affordable coverage. It should be good for folks that are eligible and enrolled in these particular plans.

Currently, 18 states have declared that they will run their own state-based exchange. Seven are planning for a state-federal partnership exchange and 26 have defaulted to federally run exchanges. So, half will run their own or a partnership while the other half will have their exchanges one by the feds independently.

I have discussed a little bit around affordability in terms of the tax credits and subsidies available to people under 400% of the federal properly level. Let's now turn the attention to adequacy -- the provision related to adequacy is called the essential health benefits -- section 1302. The ACA requires that individual and small group plans both within the exchanges and in the market place individual and small group markets cover essential health benefits. Plans covering large groups -- 100 or more employees -- and grandfathered plans are exempt, as are self-funded plans. This is an important point to make. The majority of kids with special health care needs -- we know from the surveys that the majority of kids get their insurance through large group plans or grandfather plans or self-funded plans. So, essential health benefits are going to apply within the exchanges in the individual and small group markets, but they are not going to apply to the broad population of kids with special health care needs and kids with heritable conditions. This is important to know. These are going to be helpful provisions for people eligible for this kind of coverage, but it is not universal for every single plan or policy that will be out there available on the market.

There are several requirements under the ACA with regard to the EHB that are incredibly important to be aware of. The scope of benefits has to reflect those covered in a typical employer plan, and typical is in quotes. I did that; I think it is important to be aware that we are replicating what was already in existence in the private market under the ACA in the EHB. There are some important caveats associated with that as well to help improve what is currently available. The EHB definition cannot make coverage decisions, determine reimbursement rates, establish incentive programs, or design benefits in ways that discriminate against individuals because of their age, disability, or expected length of life. There are several provisions related to antidiscrimination protection in the ACA that can be incredibly important to this population of kids. This is just one of them.

Next --

The EHB must take into account the health needs of diverse population groups, and children are explicitly identified as a population group that has to be considered. It must include benefits under 10 broad service categories which I will describe in a moment. The benefits must be

balanced among the 10 categories. For example, you can't have an essential health benefit package that has very robust physical health and [indiscernible] mental health coverage. There must be a balance among the 10 categories. Here is the list of service categories that are included under the ACA in the EHB benefits. I will not read this -- you could read it yourself. I want to draw your attention to the preventative and wellness services and chronic disease management. This is important. We will talk a little bit more about the other preventative services provisions in the ACA and how they intersect with the EHB's in a few moments.

The ACA originally called for the Secretary of Health and Human Services to determine a national standard for the duration, depth, and breadth of the essential health benefits, but after careful consideration the decision was made to use a benchmark approach so that came out in December of 2011 and was affirmed in a final rule recently. This confirmed that we are going to go with EHB benchmark plan. Instead of one standardized benefit package for all states, HHS authorized states to choose one of the following plans to use as a model or benchmark, and this replicates how the benefits were established in CHIPs several years ago. This list of 4 options available to states to choose which they wanted to go with. There were all kinds of different mechanisms that state used in thinking about what would be the most appropriate and cost effective and helpful benchmark option. This is an idea of how it shook out in terms of what states chose. 20 shows small group plans, 5 chose the largest HMO, 3 chose the state plan and none chose the federal employee health benefits plan.

There are 2 essential health benefits service categories on the list of 10 that I described that are specific to pediatrics. These are most of the time not included in benchmark plans -- pediatric vision and oral health services. They needed to be supplemented in most states. The majority of the states in terms of pediatric vision, one with the Federal employees dental and vision plan, and went with CHIP and seven already had the service included in the benchmark plan that they chose. In terms of overall health, 31 went with the federal employees plan and 19 went with CHIP and only one was included already in the benchmark plan selected. This is a resource where you can go to see where your individual state is in terms of the land they chose and the benchmark plan they chose. You can find a link to go directly to the benchmark plan so you can look at the details and it will also tell you which supplemental plan your state chose in terms of the pediatric vision and oral care.

There are a lot of questions about how the EHB's impact or intersect with state mandated benefits. This can be incredibly important to people with heritable disorders because they are designed to meet the specific needs of specific populations. In the ACA, it was required for the states to cover the benefits that go beyond the essential health benefits. A subsequent rule

clarified that, saying that state-mandated benefits that were in place before December 2011 have to be considered part of the EHB's so there will be no additional cost to the state. This was in response to concerns by states that had already developed robust states of benefits -- whether they would be penalized as a result and on the hook for additional cost. That clarification was helpful to many states. They appreciated it being offered. There was an additional clarification in the final rule that only state-mandated benefits that impact care, treatment, or services apply under the EHB's. Any limits that are in the original state-mandated benefit law still apply. So, for example, if you have a state-mandated benefit that requires coverage for a particular service but only for individual plans, that continues to exist. Any limits in the original law still exist.

Exchanges are going to be responsible for identifying the state-mandated benefits that go above and beyond the essential health benefits, as we move forward in the future, and insurers are going to be responsible for identifying the cause. With regard to state-mandated benefits, insurance plans that are not part of the exchange plans or individual or small group market -- there is no change in state-mandated benefits. There is no change in the law itself. If you have private insurance and you have access to a state mandated benefit for a particular service in a particular state, this will not change as a result of the ACA.

We will not lose anything we already had—already have.

Another thing is the Medicaid expansion. As originally envisioned, it would've required all states to allow non-disabled non-pregnant adults ages 19 through 64 to enroll. This is a new population of people. It also raised the minimum income level to 138% of the federal Poverty level for all populations new and existing. The Supreme Court said that the penalty to the states not complying is coercion. The penalty was the state losing all federal funding for the Medicaid program for the entire Medicaid program. In terms of an incentive where there are carrots and sticks -- that is a big stick. The Supreme Court said that the penalty was coercive and states could not be required to raise the minimum income eligibility or open eligibility to the new population of people. So, the expansion is still allowed, but only as a state option, not as a requirement. The states are making the choices now about whether they will expand Medicaid to this population or not. There will be 100% matching funding for covering this new recipient group in states that choose to expand in 2014, 2015, and 2016. It will gradually decreased to 90% by 2020 and thereafter. This is still higher than what is available now in terms of the federal share and the state federal partnership that finances Medicaid.

It is important to note that expanding children's Medicaid eligibility is not an option. The Supreme Court's ruling applied only to the new population of adults would previously not been eligible -- nondisabled non-parenting adults who would previously not been eligible for Medicaid regardless of their income and the overwhelming majority of states -- children are an existing eligible population so the federal government does have the authority to change the minimum income eligibility standard for them according to the Supreme Court. So, in 2014, maximum family income is going to increase to 138 Senate the federal poverty level. It is important to note that while the floor is coming up in 2014 to 138%, in states with higher income eligibility levels, that won't change. The ceiling is not changing, just the floor. We are not losing anything as a result of this provision. There is a separate provision in the ACA called maintenance of effort, which requires states to keep the eligibility on a roll, the processes that they had in place when the ACA was signed, so that people can't get thrown off a decade as a result of changes in income eligibility or administration processes.

So, what we have now is frozen in time with regard to kids' eligibility up until 2019 -- 29. -- 2019. In the meantime, nobody is losing as a result of this provision.

An additional piece of advantage is that children in separate CHIP programs -- they look like private insurance more than Medicaid. Medicaid mandates early screening and treatment which is a robust set of children's services. The kids that are currently in separate programs where they are getting coverage that is more than with higher cost sharing as a result of the mirroring of chip programs looking more like private insurance, these kids and families with income over -- under 130% will move over to Medicaid get access to reduced cost sharing and also a more robust set of benefits. This is an exciting piece as well. Currently, there are states that adhere to the federal minimums in terms of kids ages 6 to 19. The minimum for income eligibility for kids 6 to 19 is 100%. In the states with stairstep eligibility, where the older you become the lower income has to be to qualify, this has to go away and all kids across the board will be eligible for Medicaid under [indiscernible] -- under uncertainty and 38%.

A third area is cost and quality related provisions. In 2013 and 2014, [indiscernible] penetrates to primary care physicians will be increased to match the Medicare levels. It is a standard problem in the overwhelming majority of states right now. Medicaid reimbursement rates are very low for physicians. As a result, this puts pressure on providers, but also on individual families and kids because they can't get access to every provider they need. There are people who just can't accept the low reimbursement rate, and so they don't enroll in the Medicaid program and they don't accept the insurance. This is an important issue where the Medicaid payment rates are going to go up to match the Medicare levels and this will increase access for families and kids to a more

broad spectrum of providers and also help with the providers that are feeling the pinch in providing care and services to kids enrolled in Medicaid and individuals enrolled in Medicaid.

According to the Congressional budget, primary care physicians will see an additional $8.3 billion in reimbursement and the federal government is going to pay the entire cost for increased federal matching persistence to states. Providers will get more money and it is all coming from the feds. There is no cost to the states in terms of this particular rate increase.

The Massachusetts experience with healthcare reform showed us that when demand goes up and there are more people with insurance, there are more people who want to use it. So, the provider base can sometimes get stressed. The provider shortages can sometimes occur. So, in response to this concern, there is a piece of the Affordable Care Act that invest $1.5 billion between 2011 and 2015 in the national healthcare service Corps providing scholarships and loan forgiveness to primary care physicians and nurse practitioners and physicians assistants practicing in health professional shortage areas. Hopefully, this additional money into the system will help increase the providers available and decrease the risk of more people being insured, putting additional stressors on the system of provider care available now.

Accountable care organizations (ACOs) are an important piece of the ACA as well. One way to think about ACO is the individual primary care practitioner is the medical home and the ACO can be thought of as the medical home neighborhood. It is a network of medical homes linked to hospitals and specialists were accountable for cost and quality across the care continuum. One thing that is important to note with regard to ACOs is that attention is really being focused on adults with chronic illness. These are the primary cost drivers in the health care system right now. They are getting the majority of the attention. ACOs are developed in the individual states -- this would be incredibly helpful for people with clinical expertise in pediatrics and in genetics to be a part of the discussions around the development and design and operations of the ACOs to make sure that the needs of pediatrics with specific diagnoses and health care needs are included in that design.

This is an evolutionary process that we are going to see playing out over the years to come, but if we can get in early and make sure that the patient population so we are concerned with have their needs met in the design, I think this will help save us all and help achieve the aim -- more efficient and better for a broader spectrum of people.

Health homes for Medicaid enrollees is an important part. I will go into more detail in a minute.

2703 of the ACA deals with health homes. What is the difference -- we're familiar with medical home -- what is a health home? Is it the same or different? One way to think about this -- really, in the broad picture, a medical home and health home are basically the same thing. Medical home is a philosophical framework for developing -- delivering care that has several important components, including care coordination. This is also true for health homes in general. If we use the term in the context of this ACA provision, section 2703, one way to think about this is 2703 health homes are a way to fund some of the operational components in medical homes for a specifically defined group of patients that generally don't have funding available for them in either traditional or private insurance or through Medicaid.

One note I would like to make is that care coordination for pediatrics -- there is no current standard for this regardless of payer. This could make a big difference for kids and families and providers in increasing quality and reducing cost. So, I am speaking about the care coordination will happen under the section 2703 provision come but if we had a national standard that insurance companies could use and access and other payers good use and access, that could the going a long way toward providing higher-quality and less expensive care for populations in general.

Let's talk about the eligibility criteria for a health home -- state plan amended. This is just for Medicaid enrollees with two or more chronic conditions. 2703 health home plan -- at a state decided as an option to take it out -- it does not apply to private insurance, only Medicaid. People with four more chronic conditions -- one condition at the risk of developing another, or at least one serious and persistent mental health condition. How are chronic conditions defined? By statute they include mental health condition, substance abuse disorder, asthma, diabetes, heart disease, and being overweight. States can other chronic conditions to work review and approval by CMS.

What services are included? Comprehensive management, care coordination, health promotion, comprehensive transitional care from inpatient to other settings, individual and, family support, referral to social support services and the use of HIG as feasible and appropriate.

One really good piece of this provision is -- we have seen challenges in moving this forward in the pediatric and adult world because there aren't really robust funding mechanisms to support the activities that happen underneath medical home. There is an enhanced federal match for these programs. The enhanced federal reimbursement is 90% of the cost. But, it is only for health home services that I described. It is available for the first 8 fiscal years that the state plan amendment is in effect -- 2 years -- and it is okay for states to decide to implement this program. They can start with one area -- test out and learn lessons and identify high need populations and focus on them first and then spread to other areas. When they move to another area, when they open a new geographic area or a new patient population, the clock is reset and the two-year limit on funding starts over again, but only for those new enrollees under the new program. The provider types -- eligible for this enhanced reimbursement -- include a designated provider or physician, a clinical or group brassica or rural health clinic or community health Center or the community health Center, home health agency, pediatricians, OB/GYN, or other providers. A team of health professionals, including groups of this is physicians and nutritionists and social workers and behavioral health professionals—a broad spectrum of providers we are talking about in terms of eligibility.

Health teams -- specialists, nurses, pharmacists, dietitians, social workers, licensed complementary and alternative practitioners -- all different categories of providers are available for the enhanced reimbursement under this particular provision. So, that is really good. It is very broad. It should be the needs of people with these conditions in a robust and holistic way.

I want to spend some time talking about preventative services which are covered under section 2713 for people covered by new -- created after March 23, 2010, employer-sponsored or individual plans and policies, the following services have to be covered without co-pays, coinsurance, or deductibles being charged are collected.

The recommendations of the task force and the recommendations of the advisory committee on immunization practices, the Bright futures comprehensive guidelines supported by HRSA, the HRSA women's services -- some of the pieces that are relevant to genetics in this group of preventative services include screening and counseling for women at high risk for breast cancer, applies with screening, hemoglobinopathies or sickle cell screening, PKU newborn screening, autism and developmental screening, and newborn metabolic and hemoglobin screening under the Bright futures guidelines.

Last on the list but not least, are the Recommended Uniform Screening Panel -- fully insured and self-funded private plans are required to provide coverage, without cost share, in the policy year beginning on or after May 21, 2011. These have to be covered by private insurance regardless of whether the state has adopted this as a whole and CCHD and any individual condition. State health departments are not required to add these conditions to the newborn screening panels. Hospitals with CCHD or other condition screening have to be paid for by the services for patients with applicable private insurance coverage without cost-sharing. I referenced in the beginning of the time a comment regarding the importance and increasing importance of private insurance coverage as a funding mechanism for supporting newborn screening. This should be a valuable provision with regard to that. Additional screenings are important to children which are covered under other provisions in the ACA. They are included under Bright Futures and in the essential health benefits under the preventive services categories.

In summary, the ACA offers historic opportunities in terms of individual health insurance coverage and also public health, moving the public health field forward. For example, improving access to universal, continuous, and affordable health insurance coverage for individuals and increased attention to and an investment in -- real investment -- real money behind public health and primary care and prevention.

The ACA doesn't do everything for everyone. For example, the exemption to certain provisions with regard to grandfathered and self-funded plans. So, not every provision applies to every kind of public health insurance coverage. The essential health benefits are being built on existing coverage. So, some the gaps that we currently see in private insurance coverage for kids with heritable disorders—in terms of food coverage, prescription drugs, habilitation services—some of these gaps could continue. This is because the essential health benefits are built on what currently exists. We are using a structure in place now. So, there will be opportunities in the future -- the ACA was not the first healthcare reform legislation ever to be passed. And it won't be the last. If we think of as they did developmental trajectory, as we think about this as an evolutionary process, we have something in the ACA that is really good and has some important provisions in it that could make a difference to kids and families and providers and states and the federal government as well in terms of moving public health forward and increasing people's healthcare outcomes and reducing costs. There is good stuff in it, but it won't be the last thing that we ever do. There will be improvements that will be necessary to it. Both in what we fear will happen and what we project might happen in the future. There is never anything that we do on a policy level that doesn't have some kind of unintended consequence -- for good or not. We will have to be mindful of keeping an eye on what is happening with regard to the patient populations that we are concerned with and making sure that things are working for them the way that we anticipate that they will and making sure that there are opportunities for us to be

involved in trying to improve the system as it moves forward.

Long-term sustainability of state and federal funding is a significant concern in all of this. We are going through a time now of economic vulnerability for all stakeholders and patients and families and states and federal government and for providers. For all of the stakeholders involved in health insurance coverage and financing and healthcare provision. So, how it will shake out in terms of the dollars behind it is something that we all need to continue to be concerned with and we need to continue to be aware of. The need for the safety net in particular for those children with underinsurance or experiencing gaps in coverage is still going to remain critical and the ACA will not solve all the problems for everyone. Making sure that those mechanisms in place now for providing additional funding and support for things that are not covered is going to continue to remain important. In particular, state Title 5 programs and public professionals are going to have to continue to play an important role in making sure that people are protected and that the gaps are filled whatever they possibly can and when new opportunities, for improving what we have, that their expertise and influence and information is included and provided.

That was the Readers Digest condensed version. I did not cover everything in the 2,000 page document. I didn't have time for that. Probably, you don't have the time to listen to that. I want to close -- I can't resist the opportunity -- we hit the high points -- I want to mention again as they did in the beginning that the Catalyst Center is your assistance and resource for information and questions and opportunities for continued involvement in the ACA and its implications for people with heritable disorders. My contact information will be on the final slide. I can't resist the opportunity in speaking with you today to close on a personal note.

The work of all of you, as individuals and as the committee, have been incredibly important in terms of public health and in terms of individual folks who are touched by heritable disorders. I personally have a connection to this world. My daughter, Sarah, is 25 and she was born with powder Willi syndrome. That was my introduction to this world. I have been incredibly impressed over the last 25 years around the compassion and dedication of the professionals we worked with, and the way that this has constantly being looked at and evaluated, and people have been trying to improve it and working hard against oftentimes difficult obstacles in order to do that. As a family member, a person who is virtually touched I genetics and heritable disorders, I want to thank you for your work and close and open up the conversation to discussion and questions now. Thank you for all you do.

Meg, thank you for a remarkably clear and useful presentation, and a reminder to us about the fact that the safety net is important. Also, you taught us -- when each of us make health care decisions, we would like you at the table with this.

That is very flattering. Thank you very much.

With us, in each of the individual states, goes through this process. Thank you.

I will now open up for discussion. Questions and comments for the committee.

This is Charlie [last name indiscernible] Hello.

Great presentation.

I still don't understand since 2703 -- the health homes and to what extent they are currently in place and serving the needs of the children and youth and children with heritable disorders. Nurses versus other groups.

Good question. Currently there are nine states with health home spas in operation. The provision prohibits design of health home spas from concentrating on a specific age population. You can't have a health home that is dedicated specifically to pediatrics. What you can do is use other mechanisms to get at that population. For example, Rhode Island has a health home in which they did not say in the application that they would focus on pediatrics. What they said is we're going to focus our attention on a provider network which happens to be pediatric. So, the health home spot in Rhode Island has good outcome measures associated already in terms of getting additional funding and support for care coordination and for activities already happening in these centers for Medicaid-enrolled children with special health care needs of the state. Just providing additional funding and support and being able to continue to provide those good services, and expand on them. Your point that a lot of attention is being devoted to eligible -- and adults with chronic illnesses -- they are the primary cost drivers in our system. This is absolutely accurate. It is up to us to advocate with decision makers around developing health home spas that could be available for kids to meet their needs. Otherwise, I don't think people will think about kids right

off the bat. They are not that expensive to care for in the big picture. So, they don't get the time and attention that other folks do. So, with regard to pediatrics, Medicaid enrollees, I think that certainly the idea of being able to support medical homes in their efforts to provide care coordination and family education and support -- this is an opportunity to make a difference. One of the other primary benefits to this with regard to pediatrics is the way that the provision was designed, it was set up to integrate physical and mental health. That is incredibly important to a lot of kids. I do not know a child with special health care needs that doesn't have a mental health need -- whether it is diagnosable or coded is a separate topic, but a lot of people have challenges. Around physical disabilities and around special health care needs. An integration between physical and mental health can help move the whole system forward in a way that if we're looking at give more holistically, can be incredibly helpful. I think that 2703 home health spot is a good mechanism for doing that and getting additional funding support behind it. I am not sure that that answered your question.

Terrific. Thank you.

This is Dr. Kus -- this is a critical thing to look at. As Meg mentioned, most of the work has been concentrated on adults because you are supposed to show that through coordination you will reduce costs through reduced hospitalizations and emergency room. But in New York State, we are looking at it as a way to fund care coordination for children which could include children identified through newborn screening and, really, it's not funny. The one caveat -- if I got it wrong -- to me, health home is a benefit. Medical home is not the same as -- it is to enhance medical home and provide the care coordination for more, getting kids and the promise of it is that if it works with the Medicaid population, it will be exportable up to other health insurance programs. But, I look at it as a benefit to pay for care coordination for more, getting kids and complement the medical home.

Chris, I appreciate the opportunity to clarify that. There is a lot of fluidity in the way that people use medical home and health home. For me, medical home and health home are pretty much interchangeable. If you think about it as a philosophy of primary care. If you think about section 2703, health homes which are very specific and bounded by the statutory requirements under the ACA, I absolutely agree with you there. It is more about benefits and additional funding support behind it. But, as a philosophy of care, sometimes there is confusion. People use this interchangeably with medical home when we are talking specifically in the context of the ACA around the provision 2703.

Meg, this is Alexis Thompson. I thought I understood and I got confused. I am trying to understand, really, how to frame -- I am a subspecialist and I take care of kids with chronic complex illnesses. As a subspecialist, I do the majority of their care coordination. We are also strong advocates for these patients having primary care providers. When you talk about health homes versus medical homes, I am trying -- struggling as a subspecialist with this. I can see my role in both of those, but, if, in fact, the primary care is completely separate from the physical location and the reimbursement -- the place for reimbursement for services from the sub specialist, how did this come together? Is that where the health home spot comes in?

This is a good question as well. You may recall that when I was describing the integration and connection between health home and medical home, that is a philosophy of primary care practice and those terms are used interchangeably. When we talk about eight 2703 health home state plan amendment for Medicaid enrollments specifically, among the list of qualified providers eligible, are subspecialists. This not only gives us the opportunity for enhanced reimbursement, but also helps with the integration. I touched on the integration between physical and mental health, but the integration between a variety of different kinds of providers is inherent in the health home spot and for states that have elected that option to open up 2703 health home and get that enhanced reimbursement for the providers in their particular state. I think they are seeing some of that integration because it is inherent in the design. The idea of integration, in terms of the accountable care organizations, that is a different topic and we don't have enough time to get into the details right now. The medical home neighborhood -- it linked everybody. The health home spot is a weight to link everybody with the reimbursement behind it. Does that help?

Yes.

I wonder if my time is up?

We can take an additional question if needed. Any additional questions or Meg? -- For Meg?

We can open the lines for a quick public comment then or question.

-- Press star 1 to ask a question.

We have a public comment by Dean Suhr, asking about hospice care.

In reference to 2703 state plan amendment?

It doesn't say.

It is not indicated here -- it is a written comment. I don't know if you see it on the left side of your screen.

With regard to a 2703 health home spot, I've not seen anything that delineates hospital here. There is a perversion that talks about hospice care for kids with chronic conditions and life threatening illnesses. That created the opportunity for coverage without -- on a backup. Currently for hospice care, the kids have to have a life expectancy of six months or less and they need to decide -- the providers are parents -- to forgo curative treatment in order to receive hospice care.

This is Tom. -- Sometimes they get sicker faster that we expect and sometimes they bounce back and get better. With that we don't expect.

The idea that you will have a six month life expectancy that you can predict for kids with a torrential epidemiology can be challenging. Kids have different things that cause life-threatening conditions that I don't. With adults, it can be more predictable. That is a challenge. It is not just an emotional challenge for families and providers to decide that they will stop with curative care, in order to receive hospice services, but there are certain services that cross both boundaries. There are services that can be considered curative that can also have palliative effects. To say to a child with a brain tumor who can have radiation to make the tumor smaller to relieve pain and symptoms, but they can't have that anymore because they can also be considered curative, is a huge challenge in this area. So, a part of ACA that touched on hospice care took away that requirement that kids have to give up curative care in order to receive hospice services. Now they can have concurrent care -- both at the same time. Unfortunately, the six month life limiting condition limit is still there. I hope that touched on your question. If you are interested in learning more about this, the Catalyst Center has done a policy brief on financing pediatric

palliative and hospice care available on our website. If you need help finding that, please feel free to e-mail me or give me a call and I will mail it to you directly. Thank you.

Another question -- are there any provisions on medical formula and low-protein food?

With regard to those services, there is not change. The state law has not changed. With regard to dollar amount limits, they are no longer allowed in terms of services, but benefits can still be limited. So, I am afraid that -- I know that is a significant gap for many people -- especially kids with metabolic disorders. Unfortunately, there's not a lot in the ACA that touches on that. Thank you.

Another when -- what about infrastructure for subspecialist not taking primary medical home responsibility?

With regard to the state plan amendment, section 2703 -- if they are in coordination with a primary care practice, and I think the enhanced reimbursement would be allowed. Each state Medicaid program will set the mechanism of individually based on the application that they send to CMS and that it approves with regard to how the operational details will happen. If you are interested in learning more about what is happening currently, we have a list of the states that have state plan amendment -- section 2703 amendments in process now and in practice now and we can help connect you with folks to do some mentoring around what their experiences have been and how that is working in the individual states. This is so you can learn about promising practices. These have not been open for long. We don't have a huge amount of experience with them yet. But, I think there are important lessons learned that we have encountered that we could share with other folks.

Okay. Carol Greene asked this question and she is asking for the -- she understands the reimbursement, but she was more interested in the infrastructure that keeps the service available. I would imagine that is not something that the Affordable Care Act has addressed.

There are some pieces in the ACA around health information technology support. We have placed our focus on identifying the pieces that were directly related to kids and families first and more infrastructure questions second. I apologize that I can't answer that question with more

specificity right now. But, there are pieces -- Health Information Technology is a reimbursable service under 2703. So, I am not sure if that is getting to the question. If she would like to e-mail me directly, I could do research and try to figure this out more.

That is probably a better way to do that.

I guess one last thing -- I will read this -- it seems complex -- I will see if there is a quick opinion.

This is from Debbie [last name indiscernible] -- our state has thought about the possibility of families in self-insured plans buying a plan for their children with special health care needs on the exchange, assuming that it is more comprehensive. Is that a reasonable option? I realize the subsidy may not be available, depending on the employer insurance coverage.

Yes, there is a piece of the ACA around eligibility for exchange coverage that touches on minimal essential coverage. So, if you have access to minimal coverage -- meaning another insurance policy that is affordable and that is in close. Then, you're not eligible. One of the reasons is to try to keep people from leaving what is currently available in the private market and overwhelming the exchange plans and also, there are cost considerations because there are subsidies and the tax credits. If you have access to minimal essential coverage that is affordable another planner policy, then, there is no caveat that says you can't buy insurance on the exchange, but you don't get the credit then you do not get access to the subsidies which is really the piece that makes it affordable. If you think about leaving what you currently have in order to go to the exchange, you're probably thinking not just about adequacy, but affordability. So the tax credits and subsidies are the draw to that. I don't think that this will be allowable under the ACA. One piece we are struggling with -- the kids that have dual coverage. They have private insurance as an primary coverage and Medicaid and supplemental coverage. Are those kids going to be allowed to change from the expensive private insurance that they have two more affordable exchange coverages and also keep their Medicaid supplement coverage? Right now, the IRS regulations make it look like that will not be allowed. But it is hugely important for any kids with special needs in addressing underinsured. So, the IRS and CMS and a group of policy wonks like ourselves as well as advocates are going to figure out a way to make an exemption for kids with special health care needs. We will keep you posted on this.

Again, Meg, thank you for your presentation and your comments and responses to the questions. This has been very helpful for all of us. Thank you very much.

My pleasure. Thank you for the invitation. It means a lot to me.

Let's move on -- we have some updates from each of the subcommittees. We wanted to have a brief review of current and future priorities and project and where we are. The first report is from the education and training subcommittee. Don and Beth -- I am not sure who will be the presenter.

Beth can chime in and I will give a brief report.

Sounds good.

Is there a way to advance the slides?

I will do this.

The overall charge was to review existing training resources and identify gaps and make recommendations both for parents and the public as well as for a variety of health professionals. Next -- we have a committee -- five members from the SACHDNC advisory committee and a number of organizational representatives as well as other key consultants. We are looking forward to -- hopefully a time where we can be together face-to-face is a group. The first priority we have been working on this year is to identify one heritable condition that is not a part of the RUSP end for which screening and treatment would occur at a later point in child development. After that, work with professional and parent organizations to identify the major needs that would be associated with that condition. The rationale for this activity is that the advisory committee is charged with advising the Secretary about aspects of newborn and child screening. But, we are historically focused on newborn screening. We felt it would be an appropriate activity for our subcommittee to start exploring issues and opportunities around other screening. So, exploratory work is needed to understand these challenges to make national recommendations. The goal is not to come up with a condition and make a particular

recommendation for the condition, but to take some exemplary conditions and look at what the issues are that would be entailed if we did try to do some later childhood screening for that condition.

Last year, we asked for the committee members and other people to nominate some conditions. We got feedback about that process and did some more homework. The informal rating of conditions and in January, the group identified the exemplar conditions. Originally we were just going to do one, but we felt after some discussion that the different conditions evoked different kinds of consideration so we should take on more than one. For the moment, we have identified fragile X syndrome, Long QT and Wilson's disease as the preconditions. However, it recently came to our attention that a couple of years ago, there was an activity called Genetics for Early Disease Detection and Intervention (GEDDI) that we were not aware. This was between the public health genomics and genetic alliance with a similar set of goals. They also identified 3 exemplar conditions for later life screening. That report has not been issued, but there is a draft version of it and we think there are some important lessons we might be able to learn from that report. So, before we move onto the review of the preconditions we selected, we are going to review this draft -- for overlap and implications. Then, we will take this to the subcommittee in September and begin to get input from stakeholders during the fall and report our intent has been to report back to the advisory committee in the winter of 2014. I don't know if we will be complete with our activities, but we will certainly be able to give an update on lessons learned and possible next steps.

We are also providing support and input on the 2013 newborn screening awareness campaign in activities so you can skip to the next slide. This is primarily an activity led by the CDC and [indiscernible]. Our subcommittee serves as a sounding board and advisory group for these organizations as they have been planning a wide range of activities. I wanted to primarily highlight the upcoming meeting -- the 50th anniversary of newborn screening May 5-10 in Atlanta. This will be a huge exciting event and a lot of great presentations. I wanted to make sure that everyone is aware of that. The groups are also doing a variety of other activities ranging from websites, public service announcements, something in Times Square earlier this winter. Coffee table and e-books. I think it is still planned for a reception and awards ceremony in the fall and other kind of outreach initiatives. What exciting things going on in newborn screening and awareness and the 50th anniversary celebration. Our committee -- our subcommittee -- as I said -- we are providing backup and reactive support for these groups right now. But our major task will be to ask the question -- what happened after the 50th anniversary? How can we sustain the energy and not have it just be a one-time activity but something that continues to enhance awareness of newborn screening.

Next -- the third activity is to provide better guidance for advocacy groups and others regarding the nomination a review process. The project has been to collaborate with the group to develop public-friendly summaries of previously conducted reviews as well as reviews that have not gone forward. We were not trying to provide feedback to the people who already submitted conditions, but rather some high-level information for groups that might be considering submitting a nomination so that they can see what some of the that the challenges of that experience is by other groups and what kind of data the advisory committee requires before these nominations can move forward. We are trying to increase public transparency and the rationale for decisions made and support nominators and preparing successful application packages. We are working on creating short plain language summaries of evidence reviews to provide a blueprint for future nominations and also working to improve information on the website and eventually lead to some kind of lessons-learned case study book for future nominators.

One more slide -- a brief report of these activities last summer and in the fall -- last fall and spring we have been working with Atlas Research and this group has been asked by HRSA to develop draft documents for us. Now we are at a point where Alex Kemper and Beth and I are going to take the next steps on this to take the document and do more work on it and share with the subcommittee over the summer, and in the next meeting have a draft document for the advisory committee to review in the fall. I believe that is my report. Beth, anything to add?

No, I think that is perfect.

Thank you, Don and Beth. Any comments from the committee related to this presentation? Hearing none, let's go to the second report on laboratory standards and procedures -- Kellie.

A lot of these projects are being done by others. Many are on the phone. They can speak up after, if they want to add additional information.

One of the things that we have been working on last year was to -- there was CLSI a guideline -- ILA-36 Newborn Blood Spot Screening. Harry is a member of that subcommittee and he presented the draft of the document to the group in September. He encouraged comments from the subcommittee members and many people provided comments to the committee. This document completed the process and will be published soon. This will be a great help for lab, talking about developing an assay for this screening.

The two priorities that our subcommittee has -- they consist of priority A, which is a review of new and enabling and disrupting technologies as well as existing testing, and priority B, which is to provide guidance for state newborn screening programs to make decisions about implementation and integration and follow-up and quality insurance.

Part of priority B -- we have been working with the subcommittee on the newborn screening case definitions. This process started a little over two years ago. HRSA started by convening workgroups to start harmonizing the newborn screening diagnoses for surveillance and epidemiological purposes. Other milestones -- about a year ago there was a meeting with programs to review the definitions submitted by clinicians and do more editing of those definitions. And, as you can see, we have over 150 records from 16 participating states available and they are also still working on the case definitions for metabolic and hemoglobinopathy case definitions. This is being worked through these steps -- several states have been participating in pilots to beta test the case definitions. Then, I believe after the pilot test the plan is to present these to the committee for recommendation going forward.

The partner for that are the quality indicators. This is currently being led by [indiscernible]. Under two years ago we had state programs brought together to examine the Quality Indicators, and these are the states, and determine which indicators can be collected, and you try to harmonize quality indicators across the countries to gather data and be able to compare state data.

The final list has been pared down to 8 quality care measures. The idea is to move forward with these. At first, New Jersey did beta testing on one of the 10 indicators and provided impact -- they were refined down to 8 indicators. The next step is to continue to highlight test these case definitions in a select group of states. This would then -- lead to the final indicators -- then to use this for the data collection that states would collect, with steps starting in May or when a new database is up and running and ready to collect all the state data.

As I mentioned, the priority for the subcommittee is to review new technologies as well as existing testing. One of the topics brought up is the fact that for screening of Tyrosinemia type I -- the states had started using -- started using tyrosine, but it was known that SUAC was a better marker for actually measuring and screening for type I Tyrosinemia. So these doctors have been starting with a survey where the start was with 14 labs -- SUAC seven labs that used and seven

that did not in order to gather data, to understand why they used SUAC and why they hadn't considered using that marker and what obstacles there could be for them to use the marker going forward. So that we could understand why people were staying with this, rather than SUAC, moving to provide information and etc. The idea was to move on the 14th state, and get data from all states and that the CDC is analyzing this and will present the data went that is complete.

The last thing today -- under priority C -- the idea that we would like to help states when they are implementing new screening in order to -- for example -- conditions newly added. The idea is an implementation toolkit for labs, if you will. Severe Combined Immunodeficiency (SCID) as we noted here -- there is one for in process which will be helpful for labs. That is what we have today.

If you have questions, I also know that a lot of the other people from organizations doing the work are on the line. If you have questions, feel free to ask.

All right, thank you very much. Let's move on to the third report, the subcommittee on follow-up and treatment, Carol Greene.

Hi. My report is going to be even quicker, and hopefully as well organized as the last two. You already heard some of the important work from our subcommittee early this morning from Dr. Kus. You already heard about lessons learned early this morning from Dr. Kus and we really appreciate the input from the committee and from the public. You know the plans to move forward and the timeframe going forward.

This will sound very similar to a presentation we had at our last meeting because we haven't done much new work on this, that I will show you again the framework we are working on for the project on assessing outcomes for newborn screening. You will recall there was quite a lot of input from the committee and from Dr. Copeland, at the time that we spent some very intense efforts focusing this project so we would not continually confuse the committee and to make people anxious that we would be replicating work that should be done somewhere else. We are working on a framework that will allow was to assess outcomes for individuals and the public after newborn screening, and what we are looking at is the fundamental question that was asked of us by the committee, are we realizing the benefits we expect when we institute newborn screening and how do we know that? It is a two-part question. Are we realizing the benefits and

how do we know we are realizing the benefits? And to have a proper framework to answer those questions, we need to know the key questions that need to be asked, what is the data, where the data sources and what gaps are there. We're using sickle-cell as an example to make sure the framework we develop is useful but we are explicitly not answering the question for sickle-cell, but we are focusing on sickle-cell as an example to make sure we come up with a useful framework. One of our goals is somebody would be able to use the framework to answer the question for sickle-cell, but we're being careful not to tread on the toes of the experts in the sickle-cell community who could better answer those questions. Our goal is to provide a framework that would work for all conditions. Our work is in progress and I think the next slide will show you the draft framework. Thank you to those who suggested it would be less confusing if we first show a white framework and then we would also show an example of how we intend to use this to test the framework. But you can see we are looking to relate this back to the fundamental goals of newborn screening for work from the broad community, but also from this committee with the published work on the goals of newborn screening and looking at specific questions that need to be asked for each condition and specific types of measures that would be appropriate for each condition or various conditions in the data sources, and what is the key component of newborn screening addressed by each of those questions and outcomes? Again, I refer you to that paper by -- in 2011 and by data sources, these could be local, regional, state or national.

This slide shows an example for what one specific response would be. As a framework elements, one of the justifications for newborn screening for sickle-cell would be the opportunity to start prophylactic penicillin, with the goal fewer children will die preventable deaths from infection and a potential measure would be what percentage of babies with sickle-cell are prescribed penicillin by four months of age and another potential measure is what percentage of children, on age five or on penicillin, and do they get prescribed in a timely fashion and do they stay on and we look at potential data sources. In the process, we have learned a lot about limits and gaps of the data for sickle-cell and the goal would be to have a framework that would work for one disorder and bring it back to see that it works for other disorders as well.

We have already been exploring in conference calls and follow-up after the January meeting some of the work being done by other groups including groups that are supported by HRSA and the collaborative, especially in the -- group and the folks in Colorado. I have forgotten the name of the group that is. Looking at the data sources from some of the other programs and other funded resources and try to make sure we are building on the work of other groups and not trying to set up for fragmentation.

Our goal is to compare this draft framework with other existing data elements. I think it has become available now and have not been available at the time we were doing this work, but I think we have been given access to some copies and getting plans to revise the framework and we are working on this in the next week. Getting input from colleagues and stakeholders and revise the framework, and then we will bring all of this to the full committee. The next meeting was scheduled to be in September, so please expect us in September. Revised framework and a draft paper with the background and details of the rationale for the committee to look at. We don't know yet whether we are going to be asking for just input at that point or whether we will be far enough along we will be able to have sent it out in advance and have something for the committee to forward, we expect to be revised -- bring revised work.

This is the paragraph that went into the report. Looking forward to the future work of the subcommittee, continuing to work on the challenges and point of care screening that you heard about this morning, continuing to work on the framework. Before I look at the last sentence which comes back to the question about the impact of the ACA, the paper on the problems with funding of or access to medical foods is now published in genetics and medicine. We just got notification about that in the last couple of days. It is on the website, and thanks to Brad for sending that information around on listserv. The subcommittee very much appreciates the presentation today on the ACA. I think we were not the only people to ask for such a presentation but we are very interested, and I apologize I did not ask well the question our committee had during the discussion, one of the things we understand I believe also relates to impart to the question asked, on behalf of the sickle-cell community, is one of the potentially unintended consequences of the ACA as we substantially improve. I should disclose my highest that I knew it was good to start with and I am impressed with the presentation, I think we can expect improvement in reimbursement for providers and improvement in access on the part of most individuals. One of the things we are seeing in the follow-up and treatment subcommittee is the anticipation that there will be shifting in funds from funding clinics in services and subspecialty infrastructure, including salaries, to more of a reimbursement mobile, focused on the medical home and the delivery of services, even if that proves to be a shift that can be handled. There is currently not a lot of confidence about being reimbursed, and some anxiety the access to services for individuals with special needs because of genetic problems, problems with access will be compounded as funding at the state level is shifted from funding of clinical services to reimbursement. We are definitely interested in exploring that further. We know the committee is interested, in those questions, and the subcommittee is very interested in hearing from the committee as we begin to see a light at the end of the tunnel for our two ongoing projects. How to frame the question and how the subcommittee might help to address the questions of access to care as we transition to the world of the ACA. With that, I will open it up to questions.

Thank you, Carol. Questions or comments?

This is Coleen. Maybe getting a better sense of rationale, for I guess, the framework around sickle-cell and a framework you are using and the fact you are maybe not, I don't know how to say it, the benchmarks you are choosing are examples versus this might be a nice opportunity to say what those benchmarks could be based on all of the work. It feels like a little bit of a wasted opportunity in some ways and I know there are many that are getting hot, so I thought this might be a process to put into place to show that with sickle-cell disease we want to standardize follow-up somewhat from a clinical care management perspective and this could be an opportunity to show an example, but the example is real.

I would be happy to answer that. Perhaps we might want to hear from Andrea or Alexis. I should preface by saying those lines are being opened if they are not already open, that several times in the history of this project started to do just that. We ran into some issues were both people actually working on the project—we might also want to open Charlie's line as well—expressed some concern that we did not have at the table the appropriate people to make that example truly complete.

This is Alexis Thompson. If I'm understanding your question, you were sort of asking why we didn't specifically begin to address how sickle cell does or does not achieve what we hope from newborn screen follow-up, is that your question?

Perhaps I'm not stating very well. I am in the car driving. I understood her to say that as part of the project, you are identifying a handful of clinical benchmarks that should constitute, I don't know the word that is appropriate, medical care for individuals with sickle-cell disease and Carol said they are more like examples, versus what really should be done or what the consensus is or whatever. I guess I was just thinking this is a wonderful opportunity to bring people together, which we had done have done previously within the context of this advisory committee, and rather than this just being a format and a process to actually come up with those five measures, am I making myself clear?

Yes, but it is a little bit challenging, in that we were specifically told not to do that because there was the expertise and is the expertise among people on the subcommittee to look at this and provide those kinds of details. We were asked not to do that. We were asked to stick with the

framework and use sickle cell as a paradigm. It happens to be a fairly good one because several of the outcome measures we are proposing are evidence-based, and are our resources or data sources available we could access to begin to answer those. I think Carol was -- I think we were trying to make sure we were within the lines and I distinctly recall conversations, I think Sara was very much involved, where we were asked to not provide the level of detail I think you are asking for.

This is Charlie Homer, just to build on that. We started to go down that road because everyone was starting to say I think the current state-of-the-art of developing performance measures, which is kind of what you are saying, it certainly requires a lot of investment in resources [Background Noise]. The other thing was the need to coordinate with other groups. For example as you probably know, part of the legislation was to find a number of excellence and performance with Medicaid. One of those centers was charged with developing performance measures for sickle cell disease. For us to do it as one agency, entity or committee we felt we would need to coordinate with the others. I'm just sort of explaining the process. One of the questions was if we have the resources available to pull this off, and if we pulled it off, we would be to coordinate with the number of entities that were already in the process of developing these measures.

This is Carol again, thank you Alexis and Charlie, knowing there is work ongoing for sickle cell and we could certainly benefit that knowledge and our goal is to relate the question directly back to newborn screenings. Performance measures being developed for sickle cell are not necessarily being looked at in the context of newborn screening, and the specific goal for this committee to understand, among other issues for the future, when a condition is proposed to be added to newborn screening, how will we use our experience with newborn screening and follow-up and treatment, for the disease is like sickle cell for which we have been screaming for some years, how will we know if we have what we need in place to actually deliver on whatever we promised? Our goal is not just sickle cell. Our goal is to relate all of this work in sickle cell back to newborn screening and that is where we came up with the paradigm of developing a framework and to benefit from, that is why we have Charlie and Alexis and Kathy and people to directly involved in this process and on the subcommittee working on the project to bring all that back. I hope that is an answer, it is what the subcommittee was asked to do.

This is Coleen, and they appreciate the answer. I guess I am just trying to make sure that I understand all the challenges. It sounds like a lot of wonderful work is going on. [Background Noise]. It is it interagency committee here so you have that opportunity. That might it good thing. [Background Noise].

Now I will probably depart from. Here's what I understand what happened before and say on my own behalf that yes, absolutely we could be the sickle cell community, we have done that before, but I believe the sickle cell community is being convened and I personally see the bigger opportunity here as taking advantage of the incredible work being done in the sickle cell community. I think it is harder, it is harder to relate it back to newborn screening. It's a lot more nebulous and I think having this expertise focus on how we develop a framework that works for other disorders instead of sticking with one example that may or may not relate to other disorders. I think one of the things we learned in this EHDI project is we can learn a lot from EHDI but remember CCHD is different in this way. I think it is harder to try to develop a framework that works for all diseases. I think that is nebulous and difficult and tempting to do something that will give us a very concrete and hopefully useful example but in this case, I do think other people are working on the evidence-based performance measures.

This is Alexis Thompson, there should be a certain amount of caution, I think there is a framework were sickle cell, I think until it is done we should say it actually can be done. I agree, there are some groups that have been identified and charged with trying to understand quality measures for sickle cell disease. I think the real concern is even though we look to all the reasons why we should be able to apply the framework you suggested for sickle cell, I think it will be a real challenge when you try to do it to see if it can be done. I daresay if it can't be done in this disease with the extent to which we understand the evidence basis for these parameters, it will give us pause on whether or not we can do it in the diseases. I want to be optimistic but I do have some concerns it won't be as easy as we think it will be.

Thank you very much, I need to clarify one thing. What I am proposing, I think, we should not do is develop specific performance measures. We were very clear at the beginning that we were to come up with, the word framework I don't leave was used initially, but it was never intended that we would be able to carry out the collection of the data in answering the question. It was envisioned and I think it is still envisioned what this subcommittee would come up with will lead to probably like an RFA for somebody to do the work because we do not have the resources to collect all the data. That was very clear from the beginning that our goal is to set up the parameters people could use to answer the question, but we were never offered the resources to actually answer the question ourselves. I don't know if that is responsive to the point Alexis made, but I think it is clear from the important point Alexis brought out that I have not been clear and I hope that clarifies it. It was clear at the beginning we were not to answer the question, not to answer whether we are achieving the promise of newborn screening, for sickle cell or any

other disease, but we were asked to make it possible for people to answer that question if they have the funding to do so.

Carol, this is Tina. My only concern would be this group be careful and look at the other initiatives already going on across NIH and HRSA, and some of the other organizations, that have already developed frameworks similar to this for other disorders.

That is exactly why we are being very clear we are looking at this or specific purposes and looking at all of those other efforts. The goal is to harmonize. We know there are multiple efforts going on and not all of them are talking to each other and part of our goal is to bring them together. I know a lot of federal partners that work in newborn screening have regular calls where we do discuss these things. It might be to your benefit to tap into a group, and we can give you a list of that activities going on that might be helpful and useful.

That would be most helpful. Thank you. All right, I think that generated a very good discussion and I think that is going to help advance and focus what we do going forward. Are there any other questions or comments? Committee members? Organizational members? Let's open the phone lines if there are any final comments and questions from the public.

All right, if there are no other comments or questions, I think it there is no other business, I think we are ready to adjourn. I do want to thank all of you for your participation. I appreciate all of your interest in hanging by the telephone and keeping focused, when I am sure you have multiple distractions readily available in your office, so I really appreciate the efforts to contribute to the meeting throughout the day. I want to again thank the committee members, the organizational representatives and the public were attending this webinar. I want to thank HRSA in setting this up. I think you all have done a really good job in making this run smoothly and I do appreciate that. If there are no other questions or comments, I need a motion to adjourn.

So moved.

Thank you, Charlie. Second?

Second.

Thank you very much. We had very good news today about the continuing work of our committee, and you will have additional information that will come very soon about whether we will meet our deadline for a May meeting. The various subgroups that need to produce the final reports on Pompe will be prepared to go ahead with that scheduled meeting in May. Debi, do you have anything to add?

Not right now. I also want to add my thanks to all the committee members and everyone who participated in the webinar. I thought the discussions were really good.

All right, thank you and thank you all again and I hope you all have a good weekend and we will all be together soon.

[Webinar concluded]