Transcript: Thursday – September 19

Morning Session

Please stand by for real time captions.

Welcome. Thank you everyone for standing by. Speakers do have open lines today and you may speak whenever you are ready. I will be with you throughout, if you have concerns. At this time, that I could turn it over to Dr. Bocchini, Sir, you may begin.

Good morning everyone. I am Dr. Joseph Bocchini, chair of the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children. I want to welcome you all to the second meeting of this discretionary committee. I want to welcome the committee members and thank you all for coming, since this is the first opportunity to meet around the table. We appreciate you coming to this advisory committee meeting. I also thank the Organizational Representatives and welcome those here, and those participating on the webinar. I'd like to turn this over now to Debi Sarkar, the DFO for this committee.

Good morning everyone. I want to add my welcome to all of the committee members. We are thrilled to have you all here in person. The last time I checked, we had almost 200 people registered to use the webinar, to view the meeting. It's nice to see that the committee remains wildly popular. We have some great presentations lined up. I hope that will lead to thoughtful discussion and, again, welcome. I will turn it over to Dr. Bocchini, so we can get started.

Thank you, Debbie. Housekeeping notes. First of all, for those in the room here, because of the microphones, all sounds will be transmitted. Please keep side conversations and background noise to a minimum. When speaking, please state your name each time, so that we can be sure the transcript -- okay -- I guess one of the things that the operator can put in as a comment, remember that for the public -- the sound will come through your computer speakers. Please make sure the computer speakers are turned on, so that you can hear the meeting. For those of you in the room here, when speaking, please state your name each time before you make your comment. Speak clearly to ensure proper reporting for the committee transcript and for the minutes. For those who are involved in the subcommittee meetings, those of you who are here, the subcommittees will be meeting in rooms that are assigned within this building. At the proper time, you will be escorted to the room so you can get to the subcommittee meeting this afternoon. Subcommittee meetings will be from 2:30 until 4:30 p.m. Links to the subcommittee webinars, for those outside of the building, will be listed at the end of today's main committee session. The first order of business is to conduct the roll call. I'm going to go down the list for this session.

Don Bailey

Here

Jeffrey Botkin

Here

Coleen Boyle

Here

Denise Dougherty

Here

Kellie Kelm

Here

Fred Lorey

Here

Michael Lu

Here

Stephen McDonough

Here

Dietrich Matern

Here

We will not have representatives from the NIH this morning. Alexis Thompson?

Here

Catherine Wicklund

Here

Andrea Williams

Here

DFO Debi Sarkar

Here

Charlie Homer?

Charlie, you here?

Here

All right. I apologize, thank you, Charlie.

We are ready to begin administrative business. For the first item, within your agenda book, we have a response from the Secretary concerning our letter linking birth certificates to newborn screening. As you know, she referred this recommendation to the Interagency Coordinating Committee for additional review, and the response that we have is, although the ICC endorsed our objective of verifying whether or not newborn screening had occurred or expedite necessary interventions to link this information with putting the specimen number on the birth certificate, the interagency committee decided that our recommendation would be one alternative, but that there would be other alternatives, and that this would be something the states could be aware of and make their own decisions. They did not accept the recommendations that we named, but certainly endorsed the concept that the states should address this issue.

Secondly, I want to make the Committee aware that, in the future, we've made the decision that following the meeting, we are going to send a summary of each of our meetings, the issues that were discussed to the Secretary, so we can have all the activities being conducted by the Committee. I think we are aware of those. When the Committee report is finalized, I will include these reports in a letter for her information.

The third item is approval of the May meeting minutes. This will require a vote on whether or not the minutes were to be approved. First, you all received a copy of the minutes. Would like to hear if there are any additions or corrections for the minutes that were distributed prior to the webinar.

Two committee members -- if there are no additions or corrections.

With respect to content, the pseudodeficiency is consistently misspelled. I never said [Indiscernible]. I always say.

Thank you. Any other comments? [Indiscernible].

None-on our end.

Thank you. There's no additional comments from the Committee. I will accept the motion to approve. So moved, Charlie.

Second? All right. Now, we will conduct the vote. Let me know if there is anyone who wishes to abstain. Ok, Jeffrey Botkin abstains.

All right. So, now, we will go ahead and take the vote for approval of the minutes with a yes or no. Don Bailey?

Yes. I will approve.

Coleen Boyle?

Yes.

Denise Dougherty?

Yes.

Kellie Kelm?

Yes.

Fred Lorey?

Michael Lu?

Yes.

Steve McDonough?

Yes.

Dieter Matern?

Yes.

We don't have -- we have absent for the NIH, Alexis Thompson?

Yes.

Catherine Wicklund and Andrea Williams?

Yes.

Thank you.

We will conclude the administrative business. The next item is the presentation. Assessing the impact of the committee's recommendations on long term follow-up on state newborn screening programs. The presenter is Beth Tarini, the organizational representative on the American Academy of Pediatrics. Dr.Tarini is assistant Professor in the Child Health Evaluation and Research unit and the Division of General Pediatrics at the University of Michigan. Her research focuses on optimizing the use of genetic testing technology in pediatrics. She studies the organizational delivery of healthcare services through population-based screening programs, such as newborn screening, and conducts research on provider communication and decision-making about genetic testing. Beth? Are you ready to take over?

Thank you Doctor Bocchini. Thank you for funding this work. We can switch over to the slides for uploading, so we don't have that delay. Next slide? I'm sorry, could go back one slide? I'd like to thank and acknowledge a member of my research team, Shelby Lemke, who is here, was a tremendous help in developing this survey, while fielding it and helping me with the data collection. So, next slide. This project was funded by HRSA to look at, as Doctor Bocchini mentioned, the impact of the statement on long term follow-up that the committee released. So, just to provide a little bit of background to what I mean by long-term health and what we meant when we do this project, long-term follow-up is defined as beginning after birth with newborn diagnostic confirmation — confirmation of the disorder. Not to confuse with short-term. Next slide. And what do we mean for this project when we are talking about long-term follow-up? It can also view broadly. So, for what activities are involved? For this project, we took from Dr. Kemper's article on this issue and we focused on those activities that include assurance and provision of quality chronic disease management, condition-specific treatment and age-appropriate, in event of care throughout the lifespan of individuals for the conditions included in newborn screening. Next slide. The exact statement that we are looking at comes from the Committee's guidance on long-term follow-up that resulted from the meeting on September 23, 2009, when the Follow-up and Treatment Subcommittee of the main Committee convened a workshop entitled *Overarching questions, Long-Term Follow-up and Treatment in Newborn Screening*. Next slide. From that workshop came forth this paper by Doctor Hinton and others. What questions should newborn screening follow-up be able to answer, a statement of the Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children, that was published in *Genetics and Medicine* in October 2011. Next slide.

As part of that statement, there was a focus on this. This ought to identify the most important questions and issues used to inform the development of a newborn screening long-term follow-up data system. The questions and issues presented in this report guide the activities leading to the quality measures by which programs can evolve and improve. It was upon the basis of this

focus that -- next slide. Given the guidance from the committee, to examine the current status of long-term follow-up data connection activities and state newborn screening programs. This is part of a larger project to examine the policy impact of non-RUSP recommendations and other projects that are presented, preliminary results related to the Sickle Cell Trait recommendation. This is not up for vote. This issue is presented to the Committee to inform them of the work that was done, funded by HRSA and Genetic Alliance. Next slide, methods. This was a web-based survey design and it was targeted to the newborn screening follow directives. However, we anticipated that, as newborn screening is a complex system and each state is different, that simply asking only the follow-up direct would not provide us with adequate information. So, it's a snowball sampling method in which the follow-up directors were encouraged to provide us with the contact information of other participants, either in place of themselves or in addition to themselves to answer the questions we were posing. Next slide. This data collection took place between July and August 2013. There were three surveys. Respondents were supplied the opportunity to verify the data after selection was completed. The survey instrument had three main categories. They focused on long-term follow-up selection activities, barriers to long-term follow-up and general attitudes. The survey instrument was developed based on the guiding principles of the article, which is the statement of the Committee. Also, based on specific issues of long-term follow-up raised by Doctor Hoff and his papers and studies in 2006. He provided an additional statistic -- specific guidance for us on what metrics and issues we would be looking at.

Next slide. I believe we now can switch to the other presentation. Does anyone have any questions about the background or the methodology? Or the rationale for the study?

We are in the process right now of uploading the latest slides. So it will be, at most, two minutes.

Beth? Are we on the results?

I'd like to see the methods. If you can go to the results slide, it would be perfect.

Okay. Just give us a minute.

Thank you.

Can you see that now? The methods slide? Beth?

Yes, we can see it.

Great. Please continue.

Okay.

Next slide. Next slide. Next slide. Okay. We are getting there.

There it is.

Please share my screen.

I think this will become clearer later on. I'm wondering what the interface -- interface is between that and the paper the Committee has now been working on to developing measures to look at long-term follow-up. I know the efforts have been tied. Sounds like they're trying to address the same issue. A long-term follow-up [Indiscernible-low volume].

Is that an effort through the subcommittee?

Correct.

Before we embarked on this project, I had a phone conference with [Indiscernible-low volume] about this project. They had input into what questions they were asking, what metrics they were using, as did [Indiscernible], to try to reduce redundancy. [Indiscernible-low volume]

That is it.

Okay.

Thank you.

Is that the right one?

This is that.

We are going to be sharing our screen.

That's fine.

Let us know if you want to move onto the next slide. We are doing it down here on the tenth floor. Thank you.

Can you drag that screen bigger? In the corner?

Is that better?

Can you put it on slide show, or is that on slide show?

Better?

Better.

Wonderful. Thank you.

Next slide. Next slide. Okay. The development was guided by the statements and [Indiscernible]. Next slide.

For the results, next slide, please.

72% response rate, you will see the map of those -- of those states who responded. We may not have complete data on every state, in some cases the answer was I don't know to some, although [Indiscernible] these other states that responded. I want to take a moment to thank all the states. They were very gracious in taking the time to do this survey. They were very cooperative and very enthusiastic. Next slide.

This slide -- I just wanted to bring up -- it's a bit of a sidebar, but I think it's important. So, we have complete information, meaning every single item answered for 24 of the states. In 21 of those cases, that information was obtained with one responder. In three cases we needed more than one responder to gain that information. To show you what I think you know, this is not a survey method -- methodology that is one state, one respondent. It's a multi-respondent methodology. We've used it in the sickle cell project and it can be done. I just wanted to point that out. Next.

Next slide.

Previous slide.

Right there. So, 13 incomplete with one respondent. Now, also, some respondents I want to point out, when we had one respondent, sometimes they also went back, and that as a group, and one submitted all the information for the group. Next slide.

This is a tradition of the response. They had a number of different titles. There was program manager, the coordinator, the director, the administrator, the genetics coordinator. This is to point out -- remember -- we started up with the follow-up coordinator identified by [Indiscernible] on the website. As you can see, that was not necessarily the majority of respondents. Because each program, again, delegates responsibilities differently. It's important to realize when you do these surveys. Next slide.

So here are the results on the data collection. Next slide. What are the first questions we asked, does your program collect, access or contract out the collection of long-term follow-up (LTFU) data, defined as any data for individuals diagnosed with any disorders through newborn screening. Please select all that apply. We were intentionally broad in our question. Next slide.

We had 21 states tell us they actively collect this type of data. Seven states said they access existing data. Six states said they contract out the collection of the state and 12 states telling us they had no method of collecting this data.

You can see, here, we were asking them -- next slide -- you can see here, of the states that responded to us saying they collected data, which were 24, you didn't see that data set, I apologize. This was the next question to break out how you actually do it. It became clear to us and discussing follow-up with the key informants in different states, that simply collecting the data oneself as one program is not necessarily -- it doesn't mean you don't have the data. There are a number of ways to get that data. We actively collect -- access existing data, or contract out the collection. Next slide.

As your program uses the following article, authored by SACHDNC, to guide their development of these LTFU activities? Next slide. 39.5% said yes and 60.5% said no. When you break this down by whether or not they collected the data, you sought nearly statistical significant relationships. If one was asked the question, is this article in some way guiding the states? Well, a few of them or a minority of them have acknowledged that it's guided. Those that acknowledge it tend to be correlated with actually collecting LTFU data. Next slide.

So, we asked them what kind of LTFU data they collect, and for what purpose, and do they routinely monitor the data. Our points, here, and again, this is a starting point. We are not saying that we in the project determine what is LTFU data and this is the end. But we had to start somewhere and we had to define data using the previous publication. So, with that, a nice review of the literature, we came up with eight categories we felt were reasonable data categories to ask states whether or not they collect. My point is, it's not the only eight. We could add more, no doubt. Those eight, we felt, were strongly representative of a lot of the data that would be collected as part of the LTFU program. We asked them for what purpose, meaning, one could collect data for -- one could collect data for individual clinical care, to simply know whether or not a child is receiving the standard of care that one would want them to receive, or, alternatively, one could collect long term a few data for programmatic evaluation at look at in on a more systematic population basis. Wanted to know the difference. Peel apart what the states were using the data for. Thirdly, we felt that collecting data and storing it was not without actually monitoring it, is not actually surveillance. So, it's important to note if the date is not just collected, but if it's routinely monitored, which would be a step toward surveillance system, which I think is where the rubber hits the road and utilizing long term a few data. Next slide.

Here were the eight categories that we give them, follow-up status, patient demographics, health utilization, treatment regimen, health outcomes, patient access to services, cost, and enrollment in research studies. We gave them examples of each of these categories, and forming them -- informing them from the Hinton paper and the studies. On the right side you see the number of states collecting in each of those categories. The most common category was follow-up status. Whether the patient was continuing to come to clinic. The next was

demographics and health utilization, most rare with enrollment and research studies. Next slide.

I'm going to go through these fairly quickly. We can come back to them if you have questions. In order for you to see the data any meaningful way, we have to have eight of these versions. Just to go through, you can see that for follow-up status, most of the monitoring routinely is happening to improve clinical care, as well as to do research. That showed the program evaluation. At the bottom, you can see we asked them to think about missed appointments, lost to follow-up, these examples of follow-up status data. Next slide. Here, you see health utilization, interestingly enough, I'm a researcher. That's not what the routine monitoring is being done for. With patients and families to support programs in the majority of the instances.

Next was for monitoring and improving clinical care. Next slide.

The treatment regimen, as you might guess, most commonly monitored routinely to improve clinical care. Next slide.

Outcomes with equal distribution of clinical care, improving connecting families and program of valuation. You can see a little research happening. Next slide.

Patient access to services. Also, fairly equal distribution monitoring care. I think families services and program evaluation. Next slide.

Cost. Cost was most commonly for approving clinical care and program evaluation. This included direct medical costs and we asked them also about home cost, family cost, time associated with treatment cost and caregiver time period next slide.

Enrollment in research studies. None of our main [Indiscernible]. Next slide, only one state.

I will not tell you the states. I assured the state they would not be acknowledged individually. They will not be identified. Only in aggregate. So, when you look across data categories, most commonly you have 12, zero collecting any of these data. Those were the states that aren't collecting it here you can see, in this graph, that if a state is collecting data, they're collecting a fair amount. I want this slide to show that it seems like they are either doing it or not. You are collecting a fair amount of data or you are not. Next slide.

We ask them how they collected and stored. That's in some ways -- if you put everything on paper it's not going to be easy to monitor. Next slide.

Who asked them which methods the programmer part is used to collect the data. Next slide. These are all that apply. A fair number are using paper. Also, they're using computers, there was more method -- more than one method a lot. Verbal was also being used. We had not thought to include that, I should say, pointing that out when we had the survey development. That actually is a way in which sometimes data is transferred. Next slide.

On that slide, as they are changing it, the most common method that people selected were all three. So, to use a multitask method. Does your program collect LTFU data directly from hospitals or clinics electronic health records? This comes up in the statements. The vast majority do not, 68%. Next slide.

This has been an issue that I know HRSA is working on, in terms of working with states. With electronic healthcare record. The issues right now going on, -- does your program collect this data using a web-based portal system 47% say yes. There's actually a higher number using this. Next line. Does your program link or its partners collect LTFU data by linking with computerized databases? This slide in question came from the state of Michigan. Working with them and knowing -- expecting that one program can collect all the data, to answer a question well, is very unlikely. To have a robust valuation system, one has to access data from within the public health program. We included examples like school systems, for example. So, 37% of the respondents said they didn't link with computerized databases. I thought was promising. 47% state registries. Next slide.

We really ask some barrier questions, because on another conference call with Dr. Bocchini, about long-term follow-up is also not just about one disorder. It's about multiple disorders and it maybe that different disorders are more difficult to collect long-term follow-up with. So, it was not feasible to ask questions, in terms of respondent burden for each of the disorders. Nor was it feasible to ask it for the categories. We asked them the difficulty -- to try to see if one category of disorders is more difficult than the other in terms of selecting data. This is a heat map here. I knew you wouldn't be able to see the numbers. As the colors darken, that is where the most frequent answers are. Takes into account the entire summary of the entire table. As you can see, most of the dark coloring is to the left, as in less difficult. We asked a relative scale to try to get them to differentiate between them. We didn't see any statement as to whether or not one disorder seemed increasingly burdensome compared to the others. You will see the red boxes are in Critical Congenital Heart Disease, immunologic and lysosomal storage disease, which is not surprising, since they are the most recently added disorders to the RUSP. Next slide.

We asked them which method is used to store the data. Next slide.

Again, we had more than one, 24 states using methods including all those listed here. We have a lot of data. I'm almost done. Barriers, this is what people are most interested in. Why are states not able to collect more long-term follow-up data? Next slide.

We broke this down into different barriers, also informed by my previous work. We broke the barriers down by category. This is work process barriers. He then broke it down underneath states that do not collect LTFU data and states that to collect. This is the first slide of the barriers among those states that do not collect long-term follow-up data.

You can see that, basically, everyone here [Indiscernible]. They're highlighting nearly every barrier you can see there. They include communication between physicians and programs, specific data elements being a problem, whom the patient's medical home is. Newly added disorders is not much of a problem. Management of large computerized databases for program evaluation. Parental consent to collecting the data, regulatory requirements for data sharing and variation and LTFU activities needed for different disorders. You can't see the numbers, but the highest one is regulatory requirements for data sharing between agencies or clinics. Next slide.

Then, when you get to -- it's not surprising – states that do collect LTFU data, you see the barriers. On the right, you see the barriers that shift. They don't highlight it as difficult, surprisingly, those that do not collect the data. Here, the highest number is defining specific LTFU elements to be collected. And anecdotally, the states that tend to be the issue what mass amount of data are we supposed. We're collecting some, but we don't know if that's necessarily all or the right -- target, next slide.

Structural barriers. This is the states that do not collect LTFU data. Here, not surprisingly lack of employees to oversee LTFU activity. We have to thank Colorado. I would not have put that on the survey until I had a conversation and someone had said to me, candidly, I'm having issues with this project. You know, we be happy to collect data, but we can't actually do it. It would take an act of God to have us, given our statute, to collect this data and to engage in this activity, rather. So, that was very helpful. Next slide.

Then, structural barriers among states that do. You will see the highest tier is lack of a designated employee. Next slide.

Organizational cultural barriers, states that do not, tended to be more widely spread. The highest number you will see is among lack of clinician interest in LTFU activities. Next slide.

Among those that do, you see more widely spread. It doesn't seem to come out as a real problem. Obviously because they're actually doing -- they're actually engaging in it. Next slide. Then we asked them a few a general attitude questions. Next slide.

We asked because some states had raised this issue with us in the pretest. Does your program consider these LTFU activities to be part of its responsibility? We asked them whether or not -- we broke that answer down, whether or not they collected LTFU data. Now, surprisingly, if you are collecting the data and believe it's part of your activity. If you are not collecting the data, you do not believe it's part of your activity. So, the point of this slide is to say that we focus a lot on resources and the discussion with LTFU. I think that's important. But, there is an issue – there are resources issues and there are also will issues. And if a program believes it's not their responsibility, they may not work to improve their LTFU activities. Next slide.

Not everybody was doing this believes that it's their job?

Yes. [Indiscernible-multiple speakers]

They don't actually think it's part of their job?

Yes. In fact, it's funny, anecdotally, there are states I spoke with -- I have seen states like that. They engage in it, but we do it, but I don't know what we are really doing -- what we should be doing, how much we should be doing. Interestingly enough, I believe, perhaps, some of those states -- it's actually not been done with the primary aim of the LTFU. It's a secondary consequence of engaging in grant activity. So, I will use Michigan as an example. Michigan was highly ranked, obviously to be biased, witnesses in the audience, they were highly ranked across nearly every category, but they do not have a LTFU follow-up program, an official one. Most the conversation, when we went back to confirm the data with them, was the fact that they had this branch h ere, they got this grant here -- nearly all of it -- the organizational culture was, to get the grant. The data was a technical consequence.

This slide talks about -- among those is that it's not our responsibility, we asked who might it be? I think it's important, because they identified specialist, medical home and children with special healthcare needs programs among the most common. When I saw this, I thought the medical home, why would the medical home be charged with long-term follow-up activity? This would never occur to me. I came at this from the biased lens of someone who does large program evaluation, population-based research. This highlights the issue of what we and by long-term follow-up? Clinical long-term follow-up include clinical care on a long-term basis and enhancing that care. Or, does it include program evaluation, which is much broader data analysis.

Next slide. Next slide, thank you.

How feasible will be for your program to collect LTFU data for these categories within the next five years? So, we also thought -- it's great to have a snapshot that says what are people doing now? But, you'd like to know where do they think they are going to get to? Because, if the impression is, they are low now, but we can turn them up and they agree with that, that may not be an accurate assessment. I think this slide shows you that in all these eight categories, they are 30% in all of them are less than feasible, if the states do not feel in five years -- it's not feasible for them to collect that data in the next five years. Next slide.

When you look at those who say the same question – feasibility, a little bit more of a spread. You still haven't seen aggregation toward less feasible. That may mean that they just hit their max. They're doing all they can and can collect no more. Next slide.

Our conclusions were, about two thirds of states gather LTFU data with variation in the type of data collected. Most of these states collect it themselves, as opposed to accessing existing data or contracting out that collection, although a fair number do that. Most have not used the SACHDNC statement to divide -- to guide their development, but have engaged in long-term

follow-up data collection activities. Frequently cited barriers to collection were data sharing regulations, lack of statutory and authority and lack of designated employee. Next slide.

So the implications that we took from this project, these primary findings were that the barriers were multifaceted and a lot of focus has been on resource and money, which is no doubt important. But, there are other barriers that should be acknowledged, we feel, policy being one of them and value in an organizational culture. Also, the states may need guidance regarding the LTFU data collection, a prioritization of what the data collection type they should be engaged in. Guidance on the goal of that collection, back to this issue of programmatic evaluation versus clinical care. Finally, these LTFU issues, we began this project before, Pompe was added to the RUSP. As that happened, and we were in the midst of this project, it became clear that this issue will become increasingly important. LTFU data collection as [Indiscernible] added to the RUSP. Those disorders may not be clinically relevant when the child reaches a later age, at which point [Indiscernible] will be crucial. Next slide.

I'd like to acknowledge the state programs for their participation, my research members in the team -- in the room, collaborators Aaron Goldenberg, part of the research team with case and Jelili Ojodu was also helpful and involvement of the survey and the analysis. We have funding from HRSA and Genetic Alliance. That's it. Any questions, I'm happy to open the floor for questions.

Thank you very much. Thank you and your collaborators for doing this project and the results are really important, I think. We will absolutely talk [Indiscernible-low volume] discussion about long-term follow-up and recommendations for what needs to be considered. So, let's open this report to the committee for discussion.

Thank you. I thought that was very informative and really, extremely helpful. When question -- I have looked to the states responded, states like California and New York were not among them. I wondered whether you had broken down even your analyses by proportion of children covered, as well as the number of states?

We did not. We did not. That comes up a lot in newborn screening. You do a state-based analysis and do a population-based analysis. We could, I suppose. We know the data. We could then put that in as a variable and come back and see what are the number of children served. However, I guess that gets at the next point would be what question -- how are we trying to answer? We're trying to answer quality proven question from a state of the state engaged or are we trying to answer research and improving clinical care and understanding metrics of care, in which case numbers would not involve the states. Is that an interesting point you can -- the analysis will give on how you use the data going forward. Thank you.

In terms of trying to understand the impact and to get a sense from a national perspective on the number of children who we have information on, it's a little different from either of those. I think that is an extremely important issue. From my perspective, in terms of -- the position I sit in, it is information that is critical to be able to communicate. My boss and the bosses' boss.

Thanks Beth, bunch of questions, are states collecting individual baby level data on outcomes, and in line with that, does their monitoring activity imply an obligation to respond to the data that they are acquiring so that the same problems with follow-up either at the certain baby level, particular baby level or a clinic level, does the state actually respond to the information or how are those data used in a monitoring sense?

I do not know from the survey whether or not the state was collecting individual data for -- or group data. I suspect that that it could be all three, I don't know the answer. As far as what the state responsibility, that is beyond the scope of my project but I'm happy to allow the committee to discuss it. It does raise an interesting issue. If you look at something, it depends on what you fine. I would argue that also goes be on this. Immunization Registry, that goes to every registry that exist at the public health level that I know is -- are in many states reviewed. It's an interesting point. I don't know the answer.

This is Susan Tanksley, and I can respond from the perspective of Texas in regards to be question that was just asked. We collect information on the individual baby level, dependent upon response of the physician that has collected -- from the physicians forcing our specialists were seeing those babies. In regards to responding based upon that. Typically, we are getting back the information. It is more for monitoring purposes but it has helped us strengthen our resources.

To clarify that, where you collect the data from doesn't necessarily -- doesn't necessarily indicate whether it is individual or not. You can merge the databases at the public health level and still have individual level data without getting it from the clinic. That is one way to get individual level data and know the identity of the individual as well as the outcome in a massive database [Indiscernible].

Thank you, to your team for the report. To get a sense of the barriers to long-term follow-up by funding sources? What is the involvement of Title five programs in long-term follow-up?

I did not get a sense another layer deeper of the -- where the funding is coming from. It became clear to me that it is an important issue. When I was speaking with Michigan and this came up. I remember having a conversation saying you are among the top performers here. And in the next breath, I said all of your work is based on grant funding and so you are in shifting stands or a bit of a house of cards, because if you're grant funding goes way, your data collection may suffer and they thought as such. I don't know the answer to your question, but I do know that some of it is -- you can look very robust here and be potentially unstable going forward, in terms of your ability to collect.

This is Don Bailey, thank you again for the presentation and summary. In terms of how far down the chain the follow-up goes, is there any follow-up with families to understand families responses to the treatment system? That is really where the -- that is the ultimate question.

Can you give me an example?

Your treatment program, you won't know whether these kids are being followed up in getting treatment. And wanting to know are the states getting this follow-up data primarily from clinicians and programs and agencies or are they going down to the family level?

We did not ask for -- whether or not the parents reported data in a systematic way.

We could ask you questions all day. I am Coleen Boyle from CDC. I was trying to get a sense of the extent of information or that you know about in terms of the types of conditions collected, how often -- from the first screening event. I was trying to think of the robust nature of it. Or is it really selective on conditions? Sometimes we do ourselves a disservice by saying the state collects long-term follow-up and this is not at all being critical of states, because they have to operate within the context of the funding they have. But trying to get a sense of that quality and the extent of that information would be helpful.

We do not have that data. It became apparent to us that while it was very helpful, it would be impossible to do on the budget we had. And the ability to -- the time intensive nature to gather the data from the states, we would not be able to do it. It did become -- it was clear that because you collect it, it doesn't mean, in aggregate, it doesn't mean they're not -- the different disorders. As an example, what -- one state, when we asked for follow-up, they sent us specifically to the sickle cell program. That was the only program that was doing follow up -- activity. It is clear that is going up. But we are able to do it.

Charlie Homer, committee member. So the dimensions we're interested in comparing, one is across conditions, which Colleen Boyle just mentioned -- the others across states. Did you identify any situation, or is there a vehicle by which states are able to say this is what we are experiencing? Either competition or benchmarking, or whatever language you might want to learn, and did that come out in that survey?

It comes out -- I don't have it here, in the qualitative answers. I can tell you anecdotally that the survey itself was an intervention. In some cases. There were states, in which as we did the survey, it sparked a conversation about what are we doing and what should we be doing? And so I don't think it's gotten to the level of sharing among states. I can certainly go back and glean the individual comments that they had in my conversations with them. But it did spark additional discussions, which I think is in its infancy state.

Chris Kus, liaison member for ASTHO, to follow what Michael's question was, is an inference that block grants are different from other grants that are there because you regularly get them and one of the questions -- when we looked at how people fund newborn screening, it's a mix of things. One of the questions is how much may be the block grant, what is a partnership in doing that? That would be one of them. I know in our state we did some long-term follow-up specifically because we have a long-term follow-up grants and then we had a law passed for CCHD, which included no money, so we are struggling to figure what to do with that. I think the

second point you put up, monitoring versus clinical care, and when we did our long-term follow-up, the idea is that rather than versus, the appeal is you develop the system where there is more data for the clinician and then there is the reporting . It's actually a good reason. I think both of those in terms of trying to -- the better the data you get is people get to use it for their purposes which is helpful for your program.

That's a good point. With Janice Bachkus in the state of Michigan, this is where it gets to in a limited resource environment what is the type of data that can have the biggest bang for the buck? For instance, if they knew that collecting a certain amount -- a certain type of data would allow them to identify care which they could improve, immunizations, children with sickle cell, even if they did not do individual level data they could know that if they are hitting -- 60% immunization rate, that has flagging a quality issue that they can go back to the clinics with and get individual cases. People are -- they both will help each other.

Jeff Botkin. Want to see if I can get a better sense of what you think the kinds of questions people could answer with the data that is being collected. For those states that do collect data, it sounds like there are questions about whether children are getting the care that has been recommended, not necessarily which kids do better or worse depending on the nature of that care. Given the variability in treatment for many of the conditions, with some of the more robust systems, those kinds of questions could be answered to receive treatment and have better outcomes than kids who are receiving treatment?

Early on also in the development, we in the region are trying exactly that. To create a data system that -- a data set that collects exactly what you're saying about clinical data on children, moving forward, to see if they can identify what interventions, for instance, might be better than others. And so that is one of the hopes also, and Sue's collecting, one of the hard pieces. [Indiscernible] the same type of data. But the actual say metrics. Now you get into a third -- a third layer which is I can collected on healthcare outcomes, but if I don't collect the category within healthcare outcomes, if I don't collect in the same way, now I could still not have comparable data. We did not get a sense of that here. My larger sense is that, if I had to guess, is that we are not there yet. And the question is I think, can we use the data that exist now among these different programs and get answers from that right now in addition to trying to build these data sets that Sue was doing and trying to create robust similar metrics across states -- or across conditions. The healthcare outcomes and interventions. We think about a cohort study but you could get the data other ways. To answer your question, I suspect that is not the largest part of what is going on.

Hi. Susan Tanksley, representing APHL, and just wanted to comment that NewSTEPs and NBSTRN are in discussions about linking the two data sets in regards to long-term follow-up. They're trying to work through some of those issues, can it be done, etc. but the issues of consent needed for this sort of --.

A comment from the committee? Any additional comments from the committee or liaisons?

I think Chris would say the same and I know Charlie brought it up a little bit ago, so I think it is an appropriate time to mention that in the packets, is an ongoing effort, very drafty, looking for input from the committee, but I think there's a very close link between what Beth was asked to do and what the long-term -- what the Follow-up and Treatment Subcommittee was asked to do. And in fact, the project that we are currently working on relates directly to the findings of that study with our ongoing effort to work. And it goes back to something Alexis presented even further back, to work on what some of those metrics -- those kinds of metrics ought to be and how to work on harmony and to see where the sources of data are. And address some of those questions, such as what kind of data, quality of data, similarities.

Thank you for the comment. There's one question up there from Brad Therell, saying some states have a statutory requirement to maintain case registry. To get a feel for how many have this requirement to collect data as part of this?

We actually tried to do this. This came up as a side project as we were creating the data. When I was working with [Indiscernible] I thought if there are statutory -- in some states there are statutory restrictions on collecting it, maybe there are requirements and we started to go through the data. To go through the state statutes and the law. And it became clear quickly that it was difficult to tell. Because it is unclear -- you could do this analysis, but going at least from the law itself, it is very difficult to say how one could say that they were required by the law, the statute or law to do long-term follow-up. Because it is I believe, vague enough that you could go either way. There are very few states in which it says you must collect data -- it's just not there. And it is vague enough that for the state to say we are collecting data in a clinical record and that is our long-term policy, in the charts. We started to look at this and it became clear it would become far too complex from the laws perspective. And one could also -- additional methods to get that data but without, it could no longer become a quick side project.

Thank you. Any additional questions?

Alexis Thompson. Just a point of information. I was curious operationally when we see reports like this, how soon is this information available in a form that committee members can review at a later date?

The contract with HRSA and Genetic Alliance is that a white paper will come to the funder. And that white paper will be -- need to be available before the funding is [Indiscernible]. This data will be in written form, this in the next presentation, will be in the written form within the next 30 to 60 days. It will not be able to be circulated, except among the committee, because it would then meet -- be submitted for peer review. It can become an internal document for the committee, to review if HRSA wishes to release it, but it cannot be distributed outside of it until it is peer-reviewed and accepted hopefully.

Comment in its written form -- [Indiscernible - low volume] questions or comments? I've been reminded that I need to complete a roll call for the organizational representatives who are in

attendance either here or by phone. Before we get into the next presentation, we will go ahead and do that.

For the American Academy of family physicians, is Freddy Chen on the line?

We have Beth from the AAP. Michael Watson, from the American College of Medical Genetics?

Yes.

Mindy Saraco, from the American College of Obstetricians and Gynecologists?

Yes.

Association of Maternal and Child Health Programs, either Kate Taft or Carolyn Mullen? No Response.

Susan Tanksley is here from APHL. Chris Kus is here from the Association of State and Territorial Health Officials.

Carolyn Mullen is here.

Ok, great. We see you are here. Ok.

Then, Department of Defense, Adam Kanis?

Yes, here.

Natasha's here from Genetic Alliance. Ed McCabe from March of Dimes?

I'm here. I will have a conflict and won't be on the call after the middle of the afternoon but we will have someone on the call from the March of Dimes.

Ok. Thank you, we appreciate that.

Cate Vockley is here representing the National Society of Genetic Counselors. Carole Green is here from Society of Inherited Metabolic Disorders. Thank you.

Next item on the agenda is Natasha Bonhomme from the Genetic Alliance. She is going to present on the Newborn Screening Clearinghouse, Baby's First Test, congenital heart disease videos. Natasha Bonhomme has worked to improve the state of newborn screening for the past seven years, and for the past three years has overseen maternal and child health initiatives, with a particular focus on bringing the family's perspective into newborn screening policy. As Vice President of Genetic Alliance, she has launched a nation's center for newborn screening, known as Baby's First Test. We will turn this over to Natasha. I will let you begin.

Thank you so much. I would like to thank the committee for taking the time to present this update on some of our current activities. Before I dive into this presentation, I want to thank Elizabeth Bradshaw and also Dr. Gerald Martin from Children's National Medical Center, who spearheaded this program from their side. Next slide please. To give you some background, Children's National Medical Center received a Baby's First Test challenge award in 2012, to create these heart smart videos. And the idea was to address the fact that -- recently CCHD had been added to the RUSP, and it was clear there is a need for resources. They created videos, one video for parents and one video for providers and those -- children's national and Genetic Alliance, and Baby's First Test worked on the script and the filming of the videos together. I will be showing the parent video at the end of this presentation. I know some people are familiar with the Challenger awards, but the goal of the award was to address gaps in education and to improve the newborn screening process at both the community and national level. We look for -- [Indiscernible]. [Indiscernible - low volume – battery failed on microphone].

The programs that we support through the Challenger awards are meant to improve newborn screening at both the community and national level. Even if the programs are done at the community level, they always see how they can be translated to the national level. Next slide please.

So for the parent video. The provider video I will speak about in a bit. The parent video is about six minutes long. It really focuses on what parents may see if their child is getting training for the CCHD timing of the screening, on the hand and on the foot, what exactly it is screened for. We also do talk about what happens if there is an abnormal result in, and what would be some of the next steps. We emphasize the goal of screening is to detect, so there are multiple screens done to be able to detect if there are any issues. We also discussed symptoms so that even if a parent or a baby goes through Pulse Ox screening -- if they go home and this is concerned they should call their doctor. If something is off, they should not think, I shouldn't worry about that. They should do some sort of follow-up. Next slide. The provider video is 12 minutes long and that is a bit more of a training, and it goes into much more detail about screening. It is targeted to health providers and professionals and it is something that can be shown to -- even the decision-makers in the hospital. We go into details about what is CCHD, we talk about the changes in the heart structure and also the potential complication, trying to have more of a training perspective. We talk about policies, we get a history of the screening for CCHD, the decision that this committee made to add it to the RUSP. And go into a bit more detail, and said that is one, it is part of the newborn screening. Next slide. We go into details in terms of how is it done, what exactly the screen is for. There is more technical information and we also discuss how the screening works in conjunction with other screening programs. So that someone is who in a hospital setting and making a decision around CCHD or the decision-making conversations can make a better context of CCHD, in the context of -- these other types of screening. We also have a family story. And the family stories highlighted in the parent video but we go into more detail. Again, to give a more well-rounded picture of CCHD. We also have talking points -- to educate parents. That is one thing that we have found, whenever we are talking to health professionals about newborn screening, a lot of times they don't know what

words to use when they are talking to a parent. We give them talking points that they refer to when talking with parents. Next slide please. In year one, that was 2012, we pushed out the videos to a number of different -- including ground rounds at different hospitals. We were getting a number of international requests. Elizabeth and Dr. Martin really traveled quite a bit, particularly to the Middle East, because there was such a high demand of the video, about screening for CCHD. Next slide please. Because of that, year two, the beginning of this year we decided to do a focus on translating the videos. We just focused on translating the parent video because it was shorter, and it does a nice job of highlighting most of the information. And that we translated it into the five languages that are below: Arabic, Chinese, French, Russian, Spanish. Next slide please.

With all the translations, those videos were available starting July 13. They can be found in a number of different places, on BabysFirstTest.org, under our CCHD page, they can be found at children's national, under their CCHD page, also on both organizations YouTube pages. And it is also available on babies first test video page. We also printed out a number of DVDs that we are disseminating free of charge, mostly for the parent videos, but we do have a certain number of DVDs for the health professionals. We have encouraged -- we encourage people to go to the YouTube page and even if they wanted to, copy it onto their own DVDs, we really wanted to lower the barrier to using the DVDs, so they go to our website, some people can, some people just go onto YouTube and do a copy. We brought down the barrier to using the videos, but harder to track. In terms of some of the data we have, on all of the YouTube pages, the videos have been viewed over 7000 times. We are happy about that and are hoping that people are able to view the videos and that way. DVDs have been requested by a number of states. And we are still getting those in. We're running close to lunch, we will play the video now.

John, could you queue up the video please?

Give me one second.

Why we are queuing up the video, are there any questions?

Are you going to do translations for the providers?

To repeat the question, if we are going to translate the provider video. That is something we have discussed. I think that is something that is dependent on if children's national is interested in doing that, then we would be more than happy to do it. I think particularly after we do the translation of -- I think people became aware of these videos after we did the translation. Because of that, now we're getting a better sense of what has been needed and what people are interested in.

Jeff Botkin. Do you have recommendations on how the video would be used? Are you promoting this for prenatal care or perinatal care or specific time period for [inaudible]

We advocate for people to use it, the earlier the better, so during prenatal care if people want to. We have had some -- I have contact from one OB/GYN office saying they would be interested in showing it. Another piece, in terms of the dissemination, that I forgot to mention is that the newborn Channel, which works with over 1,000 hospitals in the U.S., has decided to include this in their rotation. So we're really excited about that. I am anxious to start, and it should hit their airways October 1. They're doing some editing but they are planning on showing both English and Spanish along their channels. That's a way, another way in the hospital that they would be able to see it.

This is Dr. Bocchini. Have you gone to the primary care organizations and AAFP to determine if this could be added to their parent websites or information and perhaps approval or endorsement by those organizations update?

We have not gone that far yet. I think that is a really good suggestion. We have mainly sent it out to a number of people to say this is here. Because we are getting so much -- well received, I think that is a next step.

Denise?

I was going to ask about some kind of formal evaluation of the effects of the video.

That would be great. Depending on if Children's National Medical Center came to us and wanted to do that, we would be interested in partnering with them on that. They have had a bit of turnover recently, so it's making sure there is that champion there. I know Dr. Martin is the champion, but he is also really running the whole center there. I think that would be really interesting to see, how we would be able to do that type of an evaluation. And what would be the methodology. Really focus in on one of these hospitals, particularly now that we recently have this partnership with the newborn channel, and that happened in the last two weeks and that opens up much more opportunities to get more viewership and evaluation off of that.

Who did you get the funding from?

Babies First Test every year has a set of challenge awards that we give out, and we gave this award out to children's national.

Looking at outcomes, an excellent part of this approach. Is this ready to be broadcast?

[Video playing - indiscernible due to echo.]

I apologize for the technical difficulties that we are having. We will break for lunch and we will try to solve these issues we're having. I appreciate your patience, and we will see everyone back at 1:15. Thank you.

[The DACHDNC Webinar is on break for lunch, and will resume at 1:15 pm Eastern Time].

Afternoon Session

Good afternoon everyone. Welcome to the afternoon session, the first day of our second meeting of the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children. We need to take attendance again. We're going to go down the list.

Don Bailey?

Here.

Jeffrey Botkin?

Here.

Coleen Boyle?

Here.

Denise Dougherty?

Here.

Charlie Homer?

Here.

Kellie Kelm?

Here.

Fred Lorey?

Here.

Dr. Lu was unable to attend this afternoon session. Steve McDonough?

Here.

Dietrich Matern?

Here.

[inaudible]. Oh, that's right. [Chris DeGraw substituting for Dr. Lu] Alright, thank you.

No one from NIH here this afternoon. Alexis Thompson?

Here.

Kathy Wicklund?

Andrea Williams?

Here.

Debi Sarkar.

Here.

Organizational representatives, American Academy of Physicians, Freddy Chen?

Beth Tarini?

Here.

Michael Watson?

Here.

Mindy Saraco?

Carolyn Mullen?

Here.

Okay. Then, Susan Tanksley?

Here.

Christopher Kus?

Here.

Adam Kanis?

Here.

Natasha Bonhomme?

Here.

Ed McCabe?

I'm here.

Kate Vockley?

Here.

Carol Greene?

Here.

Thank you, all.

This next presentation is entitled Policy Impact of the Secretary's Advisory Committee Recommendations regarding Sickle Cell Trait Screening in Athletes. This is a follow-up from the previous presentation that we had at the last meeting. Again, Beth Tarini will be part of this discussion. I think we will just indicate she is the professor in the Child Health Evaluation and Research unit and the Division of Pediatrics. The second presenter is Alexis Thompson, member of

the committee. Dr. Thompson is currently Hematology Section Head at the Children's Hospital of Chicago and Professor of Pediatrics at Northwestern University Steinberg school of medicine. She's on medical advisory board for the Cooley's Anemia Foundation and the Sickle Cell Disease Association of Illinois and in her current position, Dr. Thompson is an investigator on numerous extramurally funded multicenter clinical trials, as well as her own institutional clinical study, [Indiscernible-low volume]. The session will start with presentation by Dr. Tarini.

Thank you Dr. Bocchini. This presentation, I'm going to go briskly through it. You've seen it and it may jog your memory. I'm presenting it again because we finalized the data collection. In some of my background and Dr. Thompson's overlap, you will get it again. So, not to worry. Next slide. So, this project is looking at the impact of this screening mandate, specifically, the committee's response to the screening mandate by the NCAA Division 1 Council in 2010. It was approved in August 2010 and went into effect. Next slide. The mandate for Division 1 and 2 athletes was in the pre-participation medical evaluation that it shall include a sickle cell solubility test, unless a documented result of a prior test is provided to the institution, or the prospective student athlete declined the test and signs a written release. This was the original mandate. Next slide.

This mandate was largely with the result of an incident that occurred, at Rice University, when Dale Lloyd II the second collapsed during football practice and later died. His family sued a number of entities, including the NCAA as well as the university and other individuals. As a result of their settlement of that lawsuit, there were a number of pieces of settlement. The particular one at hand of the NCAA approved a proposal to add mandatory sickle cell trait screening to the medical examination. That was in June of 2010, and then in October, the advisory committee published a recommendation. Next slide. The recommendation from the committee, I'm not going to read it, but I highlighted portions that were important. That comments were made that all individuals should have the opportunity to find out the risk for various medical disorders, including carrier status for sickle cell. Next. That evaluation and testing should take place within the medical home and should include counseling about implications of the information for the individual and assurance of privacy of genetic information. That they should receive -- the athlete should receive education on safe practices for prevention of exercise and heat related illnesses. Next.

The Secretary instructed SACHDNC to work with the Sickle Cell Association of America, and other relevant Federal agencies, etcetera, to develop guidelines and educational resources about this issue. Next slide.

The National Institutes of Health and Centers for Disease Control conduct research to understand the increased risk to those with Sickle Cell Trait, the increased risk of exercise-induced sudden death. Next slide.

Perhaps one of the most important statements, Genetic testing should not be a prerequisite for participation in sports unless it's deemed genetically necessary. Next slide.

After that statement was published, the NCAA went on to approve testing for Division 2 athletes, and then in 2013 approved it for Division 3. Division 3 mandate had a waivers stipulation as part of signing -- providing the athlete with education regarding the implications of exercising the waiver option. It should be -- the student not tested but did yet had confirmed results documented or had signed a waiver shall be provided additional education regarding the risks and impact of Sickle Cell Trait.

This is -- let me step back -- I'm not here to provoke a discussion about -- for my presentation, our discussion about the appropriateness of the NCAA mandate work nor of the response. I was tasked with looking at the impact related to that recommendation from a policy perspective, and as a result of that focus on policy impact of state programs themselves. The reason I put this slide up here is that often the athletes given this one example, information that they neither Sickle Cell Trait results. Here, you see highlighted in yellow, in the inserted text, that alerting them in California if they can get such information from the state. We see here, the example of how private – a private entity has instituted a mandate which has repercussions on newborn screening program because it is a source of information said to be used to fulfill that mandate. Next slide.

Our objective was to assess the impact of the mandate on the state newborn screening programs, specifically the demand placed on the program resources, the program it changes implemented, and the variation impact across programs. You just heard about the long-term follow-up. This issue was not raised by the

committee for a vote. It was funded by HRSA and Genetic Alliance and is being brought to the committee for comment and to make them aware of the work, next slide.

So, we did have another survey just finished in August. We did phone and written surveys of the newborn screen program. We used a snowball sampling method, because this was a multifaceted issue. We looked, specifically, to laboratory directors and personnel about their experiences. We had hoped in the beginning to scale further be on the lab and the follow-up and look at potential impact on the clinics, themselves, and the patients patient coming to clinic. Whether or not the providers and follow-up clinics were having this issue. It became clear that it was too complex an issue to look at with this contract. So, we restricted ourselves to the impact on the programmatic personnel, themselves. It was considered complete after speaking for the laboratory follow-up representative and we followed each state's respondents. Next slide.

We assess the history and procedure of newborn screening for hemoglobinopathies, specifically the laboratory procedure and history, the availability of the Sickle Cell Trait results and reporting of this status. The direct effects of the mandate on the volume and nature of request and the procedure for providing requests as well as qualitative assessment of programmatic changes. Next slide.

Here are the results. Since I last spoke with, we have contacted 92% of the states and have complete information on 71%. We increase our participation since you last saw this. You will see the orange is complete response and light orange is partial and gray is no response. Next slide.

The information we found about history and procedure focused on this, while not directly related to the mandate, this was important, we felt, because you can't actually get the information about that trait unless they actually screen. Secondarily, you can't actually get the information unless they actually screen with the test that they will release and is accurate. Certainly, you can't actually get the result unless they have the capacity and resources to release it to you. Each of those pieces was looked at separately. For instance, even though state may have screened in 1975, it may not be able to provide you with result from those tests done in 1975. The years they have been screening, 1990, range from

75 to 2005 with a number of methods. Most used a two-step reflexive testing. Next slide.

This was then unforeseen but important finding. Unanticipated, I should say. There were different types of availability of result. The issue is that the assumption should not be made that just because the state screened for Sickle Cell Trait, that you can access that result. There is a part -- typology of the different ways and which the states have access to these. The top is what people presume, there is continuous and easily accessible results. The second is what we found. Some states, there was a change in data storage. While there could've been legitimate results from a previous time point, they are inaccessible because of a data storage issue. Or, they need to be transferred over. Next is inaccessible, I apologize. The next, which is disposal of records law and regulation, the state actually had to destroy the records. You have them, but by this statute, they were asked to destroy them. For periods of time, that may never allow a period of time to elapse in which an athlete at that age, in which they would require the Sickle Cell Trait for collegiate sports, would be able to access that for their own result. In some states, screening for hemoglobin started that might not have been universal. Next slide.

So, with 46 states, three had continuous and easily accessible results.21 had issues with change in data storage. Nine had inaccessible databases, seven had disposal of records due to law issues, and seven had issues restricting their ability to release results. Next slide.

Sometimes it's good, sometimes it's bad. It depends on which side of the fence you sit on. The state currently providing results of those surveyed were 31. Next slide.

We asked the state, what information did you provide? In many cases, the states surveyed provided the entire newborn screening results, 59%. 31% had just the sickle cell results. The presumption they're only giving sickle cell results should not be taken. Next slide.

Who was able to request these results? The vast majority of states a lot of primary care physician to do so. Some allow the student. Some allow the team

physician. Some allow the athletic department. A small fraction, the NCAA themselves. Next slide.

To the states who provide the results. We asked them, you allow it, now, what did you actually -- you know -- who did you provide to? What was the action? Nearly half said they provided to the primary care physician. Interestingly enough, 21% said they provided it to the team physician. 9% to the athletic apartment, and none to the NCAA, itself.

Concerns that prevented programs from presiding -- providing results, privacy of genetic information, the program policy itself, the cost, the accuracy of matching the record to the individual, accuracy of diagnosis itself, depending on the method employed to screen for trades, to screen for disease and whether trait with the secondary finding they did not feel comfortable releasing without the primary target of the testing. Results for athletes this age did not exist, inconvenience retrieving results, and use of resources and use for other things. Next slide.

Qualitative concerns that came up and we did a qualitative summary. Here are two examples. One was a value statement. Providing Sickle Cell Trait results is not a worthwhile public health initiative. This is not the mission of the newborn screening program. The second was interesting. Public trust in the program would be undermined if people found out that we were sharing information collected when you were a newborn. Next slide.

What was the effect of the mandate, itself? In terms of volume, we did this study, per year, we asked what was your annual request rate. It ranged tremendously from zero to 6,000 requests. When I sit 6,000 requests, that doesn't mean 6,000 phone calls. That could mean one batch of a request with 100 athletes on it. It doesn't mean they picked up the phone and said 6000. It does mean 6,000 results. Most requests received between May and August. This does not include record retrieval through web-based portals, if the state had a portal the individual could go to themselves, or the primary physician, to get these results. These were direct contacts to the program. Most programs had between one and 100 request that that time and not as I mentioned, it was seasonal.

We asked them about burden, and whether or not the states that were surveyed had burdens. 64% reported no burden. Of those who reported burden, 21% had only time and 15% had money. Time is always a burden and money is an additional burden on top of it. There was one anecdotal report from the state that said, was something along the lines of, we could fill an entire FTE (full time employee) just to provide this service. Next slide.

We would hire someone just to handle these calls, that we don't have the resources. Since our system is fax-based, we are killing our fax machines. We don't have funds to buy office equipment. Timewise, all of the requests come in a narrow time period in early summer, so it's like cramming 40 weeks of work into a 25 week window. Providing information to add to one of our newborns is in treatment of the higher priority than this and that is where we try to spend our time period prioritization of limited resources. Also the issue of education came up. We spend time planning to parents with the screening is for and why they are being screen, why required to get this information, I could have a tape recorder that explains where to go to get their results because I have to give that speech so often. They are getting the information, the resources and time, it's a lot of resources when the state actually does it. Ironically speaking, if the state -- it's almost like if you build it they will come phenomenon. If the state had an ability -- does not have the ability to give the results, their burden would be low because they can't actually provide the results. Of the state has the potential to give the results back, they may be experiencing extremely high burden. Next slide.

We asked for programmatic changes. They said it affected the procedure for reporting results. One said they had to make a new form. It affected their policies on releasing information. Issues about retention of results, and one state evoked a debate to destroy samples older than five years. The review of educational materials and staffing changes. Next slide.

It generated additional discussion about IT changes, whether it actually helped one state make the case for an online portal system. It brought up the issue of what if this would happen to other disorders, not just sickle cell. What should we be doing about these records for other disorders? Should we have an age limit? How long should be keep them, with the implications on funding and staffing of this? Next slide.

In conclusion, we found that not all states are capable of providing Sickle Cell Trait results to student athletes. States have varying practices for sharing those results if they do share them. Those states that are willing and able to give the results have reported variable impact of this mandate, ranging from none to significant. Next slide.

I'd like to knowledge the state programs and members of my research team, collaborators and funding from HRSA and Genetic Alliance.

Thank you, Beth. Any quick questions before we turn this over to Alexis? Okay. Let's start off with Natasha.

This is a clarification of one of the slides. Maybe I didn't see it. When you said student, was there a separate line for parent request? Or with student and parent request put --

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[Indiscernible-low volume]

Carol Greene, SIMD. You mentioned, but I don't think I saw any data relevant to it — the issue of the quality of the testing. Because, I did see some chatter on some of the listservs, about the method used and whether it's accurate. Also, I think you mentioned and didn't have data on the security. How do you know that the person asking -- because, some systems will give information to a primary care provider, but if anybody else wants it, it goes up to the attorneys and you need to (get the request) notarized and approve that you are who you are. We haven't had -- we have instances of people trying to get somebody else's data. Any data with respect to that? Or just comments?

We didn't assess whether or not the methods they use for the results they were returning, what they were. We relied on the state to say, for instance, one state said yes — I had the result from X years for these athletes. But, I'm not going to return them, because we don't feel confident in the method that was used or in the fact that the test was not confirmed. So, we did not assess that. We allow the program to make that assessment and give it to us. We didn't ask specifically what the process was, the identity of the individual. Anecdotally, with the conversation with the states, there were pages where the team physician would qualify -- not an identity issue, but a labeling issue -- what is your primary care

physician, if the physician qualified under the statute for the program information. It became clear that the statutes, obviously, worked built for this situation. There are issues that can come up that you would be -- would be unintended or unanticipated.

Alexis?

This is a perfect segue. You will see there is substantial overlap. I think most of what I'm hoping to do is to frame some of the policy issues that have come up across other stakeholders that may be worth reflecting, as well as some of the unintended consequences of the mandate. Going to go work with some of the slide that overlap with the ones that Dr. Tarini just presented. Next slide.

We've already reviewed the reports. I won't review that. There have been some interim events. I think that this has given us quite a framework for the impact on states, but I think it is certainly, at this point, a question on whether or not the Sickle Cell Trait notification has broader implication across other inherited conditions, which is already been discussed today, as well as proposing next steps for discussion by the broader committee. Next slide.

I think, again, that's always worth going back to the bedrock. That is, newborn screening saves lives for children with Sickle Cell Disease, as was the intent. We know that families, who noted that their children have Sickle Cell Disease from the newborn period, are more likely to survive, compared to parents who determined it any time later. Certainly, the landmark study by Dr. Gaston and her colleagues in the mid-80s that showed there was an 86% reduction, and this randomized clinical trial for children who are receiving penicillin, compared to those who had not, an 86% reduction of risk of a pneumococcal infection, lead to a consensus conference by NIH — simple reliable tests to justify mass screening. Fast forward to more recently, data from the Dallas cohort and other large states, would show that 98% of children with sickle cell disease will survive to adulthood.

This slide looks like it got tipped a little bit. This was just a note for the range in terms of dates for universal screening. The earliest being New York in 1975, the most recent being New Hampshire in May of 2006. The next slide breaks down in an easier way, looking at how many states were covered, when. By 1990, about 40% of states had gone on to have -- to include Sickle Cell Disease in their

universal screening. So, this would have been in the period just following the data that I just presented. It's worth noting that by 1995, 76% of states initiated newborn screening for Sickle Cell Disease and it's an important time point because children born in 1995 will be 18 this year. This actually would be the cohort for which this is most relevant. When we look at the states that were included, really, I think the only state that had a substantial population and likely a population of risk or sickle cell that would not have been included, at this point, was the only one outstanding, was Georgia. Certainly not screening at this point in time period, the last of the largest states with diverse populations were certainly included in that 76%. Next slide.

When we think about the NCAA, if we can scroll through these, I think that's had a recover these. The mandate in 2010 for solubility testing, which I will touch on, how it arose and the next one. There was opt out for prior testing, or they're willing to waive liability for the University and the NCAA. Next slide. Then, that had already been mentioned, the expansion to Divisions 2 and 3 with the change in the Division 3 vote. Next slide.

We've already gone over what the Secretary's Advisory Committee recommendations were. I should mention these were the ones accepted by Secretary. The ones recommending actions by the CDC and NIH were not accepted. Next slide. I think it's worth noting that there are a number of other organizations who have weighed in on this. The following year, the Sickle Cell Association of America published their perspective. The American Society of Hematology in 2012, which was endorsed by the American Society of Pediatrics, American College of Medical Genetics, the American Public Health Laboratories-- and the clinical pathologists have all endorsed the ACH statement which I will present in a moment. The American College of Sports Medicine, which includes team positions, also has a policy as does the American Academy of Pediatrics. I will present that one, as well. On the next slide is the position of the American Society of Hematology. Just for full disclosure, I am a board member. ASH does not support testing or disclosure of Sickle Cell Trait status as a prerequisite for participation in athletic participation. Instead, they recommend implementation of universal interventions to reduce exertion-related injuries and deaths, says this approach can be effective for all athletes, irrespective of their sickle cell status. It's very much in keeping with the policy position of the US Armed Forces. ASH believes the NCAA policy, currently written and implemented, has the potential to

harm the student athlete and the larger community of individuals with Sickle Cell Trait, and ASH strongly supports increased biomedical and population-based research on Sickle Cell Trait as it relates to exertional injuries as well as other clinical conditions.

The Academy of Pediatrics does not have a specific position on Sickle Cell Trait. However, it did publish a collaborative policy with the American College of Medical Genetics earlier this year. They do not support routine carrier testing in minors when such testing does not provide health benefits in childhood. They specifically advise against school-based testing or screening programs, because the school environment is unlikely to be conducive to volunteer participation, thoughtful consent, privacy, confidentiality or appropriate counseling about test results.

Next slide.

[Indiscernible-low volume]

I believe this statement is about legal minors. This is actually verbatim from their policy statement. So, in terms of fulfilling the NCAA mandate, certainly, their mandate for Division 1 using solubility test, but that methodology is problematic. It is recommended but not required. One can obtain existing results from the primary care provider. One can have their primary care provider retest, although, certainly, there are no provisions for Division 1 for cost. There have been some provisions made for pending testing, through colleges and universities, with some assistance for the costs of those tests. The NCAA actually compiled a list of all 50 states, in terms of how one might contact by phone or the web for every single state office. If you wonder why you need [Indiscernible], you can see NCAA. Next slide.

Just to go over the Sickle Cell Trait testing. What the NCAA record for solubility test, this test is of no use for primary screening, and is not used by any of the 50 states that do newborn screening. It's often negative, in the newborn period, who have high-levels of affect and negative in Hemoglobin C, the second most common form of Sickle Cell Disease. It cannot distinguish between Sickle Cell Trait and any other form of Sickle Cell Disease. Individuals with hemoglobin S or hemoglobin related Thalassemia cannot be given reliable information about their

condition status using this test. In an emergency, it might help raise the suspicions of Sickle Cell Disease. In the period prior to newborn screening, an African-American baby and the emergency room might have this test done to decide whether they would be hospitalized. In essence, that test had some use in that era. It can also help distinguish between sickle hemoglobin and other hemoglobin that had similar electrophoretic properties. For instance, if I have a patient in my office who is not in an ethnic group and I would not expect of Sickle Cell Disease, I can certainly, I have a negative electrophoresis pending, but they have a negative Hemoglobin prep, I'm are likely to think they have hemoglobin G or hemoglobin D, which electrophoretics could look like. Next slide.

What are some of the unintended consequences? We already know there's dramatic increase in requests in a summer of 2013 for newborn screening results. The practices and policies have just been described among states, it's been highly variable. Part of it is the retrieval of the results and a lot of it is policy implications and release of this information to third parties.

There are some additional unintended consequences. These are ones that clinicians are already seeing. What about high school? High school athletes who aspire to play college sports? There have definitely been states and counties already, where mandating Sickle Cell Trait results for high school athletes. Again, it may certainly point to the NCAA as their reference for requiring this. The second scenario, though, is the one I find most disturbing. That is, having the mother of a child with sickle cell disease who does not believe that she should exercise, for fear that she actually is in peril with her own health, though clearly this may be someone who has other cardiovascular risk factors, including obesity, yet is afraid to exercise because she actually perceives that risk, that the NCAA has prescribed, actually refers to her. Next slide.

What are some unresolved issues? The question is a legitimate one. If status reliably determined by the methods used for newborn screening? Without question, when states elected to choose methodologies, the focus was on disease. Certainly, in many states, if the states have a two-step program or the expectations are that a clinician actually confirms those results. The question is, what is the intent to use those tests alone to reliably determine the status of traits? Also, one of the current state practices? I think Dr. Tarini's work is giving us some sense that there is a wide variation, in terms of what the states' practices

are, in terms of whether states do have resources for that. The mechanism of retrieval. At a much later date -- clearly, there are opportunities for education and awareness of individuals and providers. It's no question about that. Reproductive choices, it's certainly one very legitimate reason for wanting to know later about one's sickle cell status, as well as, potentially, other health consequences of health carrier status. In each state the remains a question of how can a provider readily access these records. Next slide.

I think these are things that are worth the committee is considering. Do we believe that the recommendations that were published two years back, should they still stand? The question has already been raised. Is this an appropriate use of newborn screening? The NCAA is the largest -- this is the largest expansion of mass screening by a non-public health entity and, certainly, that is some that -- me personally -- I have some great concerns about it. The question is, what if any role we can have, wanting to defined some guidelines for that. Can or should the committee provide additional guidance to the Secretary or to states regarding response to these requests? How does this experience impact the broader discussion of notification of carrier status for other conditions? Next slide.

Is sickle cell exemplar for carrier testing? We know that according to Healthy People 2020, everyone should know the status. Is there limited empirical evidence of the value in actually doing testing later? We all need to understand what two people do with information about carrier status? The obligation to the child, separate from the application to the parents and the larger family, is there consensus on disclosure? This certainly is some discussion in the literature about what are our obligations. The question is, have we resolved all those issues about disclosure? That incident to be issues about the biomedical ethics considerations, in terms of respect for the autonomy of individuals to make decisions, the obligation of providing benefits and balancing benefits against risk, and the obligation of avoiding harm. Again, the logistics of public health entities, even for legitimate reasons for doing this testing, but just public health entities must consider in providing information about carrier status.

My proposed steps would be, one, depending upon our discussion today, whether or not we need to consider having an ad hoc working group to actually answer some of these outstanding questions, including whether or not it's appropriate to provide the additional feedback to the Secretary on any new concerns that have

arisen. Another is potentially to develop guidance for states on handling requests, and to envision a framework for dissemination of trait status across other inherited conditions.

A very nice summary of the issues and review of what we need to consider. Let's go ahead and open this discussion to the committee and the liaison. Remember, when speaking, please speak clearly into the microphone and give your name so we have that clearly defined in the transcript. For the public, if you wish to ask a question, please go to the lower portion of the chat box, type in your question, click the send icon, and it should appear on our screen. We will start off with the committee.

Thanks Alexis for that presentation. I was wondering whether or not there is any systematic effort to collect more data of some of the examples that you put out there from a clinical perspective? So, from a survey of clinicians and seeing people come in with the symptoms, have there been any relevant anecdotals? Do we have any systematic way to collect that kind of outcome or impact?

It's a really interesting question. To my knowledge, there has not been. There have – there's certainly a lot of anecdotal information. Many people have had similar kinds of experiences. I think, to my knowledge, there has not been much. There have been a few who have attempted to collect information from student athletes, about how this has impacted them. I would say that those that I'm aware of, have gotten limited cooperation from the NCAA.

Thank you for both these presentations. It's a fascinating and complicated issue. On the one hand, there's no scientific basis, basically, for this recommendation. Lots of groups have looked at it. Science basis suggests there is no increased risk with people for Sickle Cell Trait for sudden death, which is the rationale. That is appalling for a scientific group like ours. Second, we don't write sports groups, which really have no basis in knowledge and scientific things, making public policy recommendations which seem to have a profound impact. That was the basis for our previous recommendation. Then, the flipside, though, there is Federal legislation, as we all know, recommending that it is not only established newborn screening, but the newborn screening program, which we are the national coordination center, says we should do everything we can to notify families about trait status of individuals with sickle cell. There should be very active programs to

educate people about their trait status, that they should be educated by the implications, particularly for subsequent reproductive issues. So, there is sort of a compelling public interest for widespread awareness for trait status. So, it's an interesting tension here. We basically don't like the scientific argument of putting the big burden at the wrong time, perhaps, but on the other hand, it is serving, potentially, a different public health purpose, which is increasing -- again, we don't like that it's only in a specific population related to -- well, it's discriminatory. Based on their trait status, they are not told whether they need to be informed of the trait status. We are not saying -- I don't know -- whether we are [Indiscernible] or there is value, as you were starting to do, separating the mandate specifically around screening for athletes. The other issue is , what other strategies to enhance awareness? I'm just thinking this through out loud. Perhaps this is dangerous for a committee member [Laughter]. One of the underlying concerns is, oh my gosh, state labs are asked to provide information to the public about trait status. Newborn screening, tell me where I am wrong? The newborn screening program that we are supposedly coordinating is supposedly all about informing families about trait status. So, tell me where -- people are looking at me -- I'm just trying to provoke discussion. [Laughter]

If I can just respond to part of this. I think that it's important, that one talks about informing. Fundamentally, one must, before doing this, understand why you feel the need to be informed. We are very clear on the reason why we think families with babies who have Sickle Cell Disease are notified when they are is because they are at substantial risk of dying and it can now be prevented. We do not have that kind of information. So, when you talk about knowing your status, what else are you telling them? In the absence of science, one has to really be concerned about missed attribution of risk. I think that, fundamentally, that is the problem right now. So, to me, in my opinion, that can be as harmful, if not more harmful, than that of our current way of doing things. To the extent there is discussion about reproduction and there is a perception that we have knowledge, as you already pointed out, we do not have, about the risk for athletic participation. What is that conversation? It really makes it very difficult for a state agency, let alone a provider, to be able to frame beyond the reproductive risk, what other additional evidence-based information we provide for families or individuals at that point.

If I may simply, and again, please correct me where I'm wrong. The sickle cell disease demonstration program legislation requires us to report on the activities of states that are participating this program, about not only sickle cell disease notification, but what we've done to identify individuals at trait and what programs are being done to educate them about that.

I think that goes to what Alexis just said. When you look at the risk for individuals with trait, it's having a child with sickle cell disease. If you take the reproductive issue out of trait, then you have to look at it a little bit -- is there still a reason for us to inform individuals that they have traits? I would imagine that there is. I can't point to a study or reason to inform them. There aren't other risks besides just athletes. They are small, but they are in the risk. I think you have a couple, as Alexis was saying. The rationale for doing these screenings with how you are going to move forward to newborns and informing people about their status. We would look at it as -- we want every individual to have a medical home. You go back to your recommendations at this committee made. What's being done on that front? Regardless of the need that they have a medical home. Look at it in the context of that, and it makes the argument a little different than saying you want them to be notified [inaudible].

This is Andrea Williams. I am very well aware of the sickle cell trait follow-up care your follow-ups. It is done by genetic counselors, it is not done by state employees sitting in an office who does not have any idea on what they're giving to the person. In that cushy place of genetic counseling, proper follow-up, make sure they understand what the risks are for having a child with sickle cell disease. There's also a place for them to discuss genetic information and how to share the information and whether they want to or not. So, I think it's a totally different conversation. We would not want targeted screening, targeted screening has always been across the table for all of this. That's the reason why we don't target African-American women in the prenatal period. You're only going to screen the African-American women across the board. So, we really need to think about what we are talking about.

Kathy and then Steve and then Chris.

This is Cathy Wicklund. [Laughter] I can give it back. I was going to build on what Andrea said. The issue, too, is not even whether they talk to someone at the state

or not, but if they are getting their information back from a coach or a trainer, whether that includes any discussion about reproductive risk, and one of our students as part of her thesis project, actually surveyed several trainers as to what they were actually providing when they gave this information. Of course I can't remember exactly the result. But, it's really variable as to what they're talking about to the kids when they are giving this result back. Again, it's not set up with an infrastructure that's there to address all of these other issues that we are talking about.

Jeff Botkin. This is very interesting and I think very disturbing. [Laughter] Two questions, really, one sort of vague and one specific. Alexis, you had listed a number of organizations out there that have taken a stance on this but didn't really tell us which were supporting and which were not supporting it. Are there other professional organizations out there that are supporting the NCAA on this?

[Indiscernible-low volume]

Okay. So, the sports organizations are together on this. They are consistent contrast to all of the health professional organizations, is that the status of it?

The answer to that one, by and large, is yes. And NATA is only one that clearly in support of this. The American College of Sports Medicine is a very nuanced response. They are absolutely -- their first line indicates that they do not think that anyone should discriminate against people have Sickle Cell Trait; they should be allowed to participate fully. These are by and large team physicians, and many of whom are employed by the universities. So, certainly one has to knowledge that some of that, certainly, does create if not an absolute, certainly an appearance of conflict of interest. They have been very reluctant -- Hematology has met with them and they're looking for other -- many of them genuinely want to advocate on behalf of our student athletes. They truly do. I think, in their world, they actually thought, that they actually saw a scientific basis for it. I think many of them were actually very surprised, hematologists, for instance, looked at the same data and came away with very different impressions.

[Indiscernible-low volume]

Thank you. To make a quick, I guess, first of all, I'm tempted to encourage an ad hoc working group on this, cognizant of the fact that this isn't so much as secretarial issue to the extent that we are advising the Secretary, but the newborns and children's piece is part of our mandate, and this might well fit under the mandate to comment about genetic testing in children that might improve the status of the situation. Maybe a quick question here. Theoretically, a lot of these kids who have trait should know that, if they've been through the newborn screening program. Don't know if you got a sense of that, when you were talking to the states, that parents were calling to get verification of that so they could show it, or, is this new news to people? It's against this larger issue of whether anyone has shown utility of disclosing trait status out of newborn screening programs. I'm not aware of any literature on that. It seems to me that this committee might want to think in some detail about whether this is a useful function here, because for the most part, if indeed the value is for folks who are considering reproduction, then we ought to offer people, considering reproduction, trait testing, not relying on the newborn screening program to do reproductive counseling 20 years in advance of having a baby.

Alexis, you want to respond?

No.

We will send that down to Stephen and then Chris and then Natasha.

Do you know if the NCAA policy, if that was a part of the legal settlement with the NCAA and do they have flexibility, or are they stuck with this policy?

The NCAA has not been interested in sharing the actual information about how -- they volunteered this is what we're willing to do in response to the damage request on the settlement. The settlement is not public. The NCAA, though, has created a large number, ordinarily large number, but certainly, they have a large number of educational materials both in written form and video, for student athletes. What we know is happening in the current environment, is that they are moving toward an increased awareness of sickle cell trait. Some of their attributions, again, some of our struggle with the science behind some of the attributions, for instance, they talk about something called exertional sick link. Most of us in the field of hematology have trouble with that terminology. There's

nothing to document that sick link takes place. They point to things like, for instance, autopsy reports were you see sickle blood cells, even though, I think, Hematology one-to-one with tell you that if you are not breathing and you have sickle cell trait, no matter why you're not breathing, you have sickle cell. So, the notion that bad, in fact, defines the fact that there is process going on scientifically, is really troubling. The NCAA has established a science Institute, that just went online this summer, they say, at some point we will actually have our assays that will come out of it for research study. But, at current time, we are not aware that anyone has been funded by the NCAA to do that.

The issue of whether or not there is evidence -- the report in 87, the New England Journal showing the association between Army recruits with Sickle Cell Trait and death was the basis of this, although I understand your point about physiologically – why there's no explanation, there is a case study that shows this association. So, I'm wondering what your thoughts are about that for this issue, which was a secondary case, whether or not there is actual evidence, some evidence suggests that extreme exertion could be associated with sudden deaths in an individual Sickle Cell Trait, not to defend the NCAA's intervention because the Army's response to that was universal precaution, not screening. It's a separate issue. Is there a concern that there is a small risk in a subset of population placement place in extreme condition and secondarily, what is the response you take. I was just wondering what the Carr report place in this.

John Carr, in fact, accepted my report, done in 2010. The report looked at those branches of the military is that, in fact, have embraced universal precautions and those that have not. Certainly, those that have embraced it have seen a reduction across all individuals just in basic training and, so, it clearly is good for everyone to be able to rest, to be adequately hydrated. I dare say, it's very [Indiscernible] to say our troops are any less prepared than NCAA athletes for action. One has to balance that against harm, to the extent that one can actually reduce complications and death with relatively simple interventions, it's sort of begs the question, if one can, in fact, look at a universal approach, can one then achieve benefit without the harm that potentially comes from stigmatization, discrimination by doing testing.

[Indiscernible-low volume]

I think that the Association is certainly -- certainly those and yourself included, that, were epidemiologist, the notion about there being association and framing that in terms of causality, I think is a struggle. I think that often, at this point, frankly, that is where the difference is. I think the hope, and I hoped to present today, that there was actual impaired research that was being done to clearly define the connection. We now know that there are a number of metabolic myopathies that are disproportionately seen in African-Americans. So, one of the questions is, we have any sense of how many of these individuals, as rare as they are, do we have any idea how much of those actually have these metabolic myopathies? One could actually easily imagine that, given how common sickle cell trait is, again, some people have metabolic myopathy and have Sickle Cell Trait. These are the kinds of things that we think should be part of the dialog. There are real opportunities that are being missed.

Chris, then Carol.

Chris representing ASTHO. Test questions, one is, when states are contacted, what happens? Do they offer counseling? Do they tell them to go to the medical home? Do they get information about that? The second thing is, when the athletic department gets the information, what do they do with it? Could it be mixed information, and are the athletes than treated differently? Are there different programs for them? What we know about that?

It depends on the state. If the request comes directly from the individual, if the request comes from the University, the team physician, they will not necessarily have the access to physicians for the individual. In a few cases we spoke with states who said they had tried, reached out to say we will help you. We will release this information if you will let us help devise an education strategy, and they were rebuffed. To your point about inside the institutions, it's very difficult. [Indiscernible] … grant has worked on this to figure out what's going on.

Natasha, Carol, Andrea, and then we need to close the discussion at that point because of the time frame.

Thank you very much. A comment and a question. For the comment, not only were states getting an increase in requests around Sickle Cell Trait status, but the newborn screening clearinghouse also got a number of requests asking how do

we find out this information? Who at this this stage should I be calling? For us, what we did was -- I would contact the state first and say, how would you like me to handle it? A lot of times they were saying, just as the data shows, we don't have this data anymore. We can't get it out. We can't access it here. Tell them to go to their Pediatrician. One thing that I am looking to do, would be to contact those parents to say, did you ever get an answer? Because that is the concern of mine. Parents are going -- students still don't know kind of what the roadmap. That also is going to look poorly on the public health system. Most of those requesting, as Dr. Botkin suggested, it was new news. It was not, I would like to confirm this. It was, how do I find out what the status is? I think those are all very important pieces to think about as we move forward. Then my question is, since the advisory committee report on this-- what is the communication with organizations that were listed with the NCAA or the [Indiscernible] around this. I'm guessing not much, or the NCAA may have not been an active dialogue, but maybe I'm wrong. Also, has any group reached out to the family that was involved with the lawsuit? I think that, coming from a sidebar with Susan over here, that they may be more open to thinking about the different opportunities for really addressing what their core issue is. I don't think their core issue was, potentially, being discriminatory against these student athletes.

Carol?

Several things, ending up with coming back to one of Alexis' questions. But, it seems like sickle cell is frequently -- it's ahead of everybody else in bring up interesting end up working questions that are going to apply to other disorders. One thing I would say, for sickle cell, one of the things that's been drilled into me, is adults often know their carrier status but not always. So, one of the reasons that it's important, and I think we've heard this from a member of the committee before, one of the reasons it's important to tell families about the carriers, the trait status of children, is that the parents may have never been offered or have declined carrier testing and might not know, until they learn that a child is a carrier, that they are at risk with having an infected child. It is not the purpose of newborn screening, but it's been used [Indiscernible]. With that said, there sometimes issues of paternity. That would be another one, when the 18-year-old knows that it's just a matter of formality, he's got to show the piece of paper to his team, because he can't possibly be a carrier, because neither of his parents are carriers, because they have this dining room talk and oops, his daddy is not his

daddy. That could be some other -- anyway, I'm interested. I did not know there was a difference in ancestral populations of some of the metabolic myopathy these, what happens when we are doing -- doing whole [Indiscernible]. What are we going to tell people when they are a carrier for [Indiscernible] or a carrier for Pompe or a carrier for [Indiscernible]. All of those are some of the common reasons why people get [Indiscernible]. We see that when we are treating the military. What is this all going to mean for what we tell people about, if it's going to be treated -- what's the word -- an incidental finding, or should you really be telling people that they're carriers for some of these things because they actually might have, probably more medical implications than Sickle Cell Trait?

With that, I think we've had enough discussion to make it clear that this is not only an important issue, but one that the committee should be involved in. I think the authority made the recommendation that we adopt the recommendation for an ad hoc working group. I believe we need one. Unless I hear something from the committee against that, we will go ahead and set up an ad hoc working group to address the issues, in a way that Alexis so nicely put together, and bring that back to the committee for discussion. I think Alexis is going to be involved. Are there any other people? [Indiscernible] Natasha, [Indiscernible] Carol Greene, Natasha, Wicklund and Cate. Okay. Thank you.

This will conclude the Thursday session of the main committee meeting, and subcommittees meeting will begin shortly. In view of the hour, let's say they will begin at 2:40 pm Eastern time so, for members of the public, if you do not wish to attend a subcommittee meeting, please be sure you close your Internet browser window so you can be logged out of the webinar. If you wish to attend one of the subcommittee meetings, beginning at 2:40 pm, please click on the subcommittee link, displayed in your webinar window for the subcommittee you wish to participate with. To avoid audio problems, please do not log into multiple subcommittee webinars at the same time. Also, remember to keep your computer speakers turned on, for that's how the sound comes through. With that, we will conclude this session and each of the subcommittees will meet and begin. Thank you.

[The DACHDNC Webinar has concluded Thursday's session, and will meet again at 10:00 am Eastern Time tomorrow, Friday, September 20].