25TH MEETING OF THE SECRETARY'S ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN

RENAISSANCE WASHINGTON, D.C., DUPONT CIRCLE HOTEL

SEPTEMBER 22-23, 2011

COMMITTEE MEMBERS PRESENT:

R. RODNEY HOWELL, Chairperson

DON BAILEY

JOSEPH A. BOCCHINI, JR.

JEFFREY BOTKIN

REBECCA H. BUCKLEY

BRUCE NEDROW CALONGE

FRED LOREY

ALEXIS THOMPSON

TRACY L. TROTTER

GERARD VOCKLEY

CHARLES HOMER

STEVEN McDONOUGH

CATHY WICKLUND

ANDREA WILLIAMS

DIETERICH MATERN

EX-OFFICIO MEMBERS PRESENT:

COLEEN A. BOYLE

ALTERNATES:

CARLA CUTHBERT

DENISE DOUGHERTY

KELLIE B. KELM

SARAH R. LINDE-FEUCHT

SARA COPELAND, Secretary

ORGANIZATION REPRESENTATIVES:

FREDERICK M. CHEN

MICHAEL S. WATSON

JANE P. GETCHELL

CHRISTOPHER KUS

BENNETT LAVENSTEIN

MARY J.H. WILLIS

SHARON F. TERRY

ALAN R. FLEISCHMAN

CAROL GREENE

DR. HOWELL: Ladies and gentlemen, let me welcome you to the 25th Meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. This is a very unique meeting in the fact that we have a considerable transition within the committee this time with the considerable number of folks going and coming. Let me first comment about the new members of the committee, who we're very excited to have outstanding new persons coming on the committee.

The members have copies of the CDs of these folks, and so, I'll be fairly brief. But the first comment that I'll make is about Dr. Charles Homer. And I don't know whether he's here or not.

I haven't seen him. Have you?

MALE SPEAKER: Yeah.

DR. HOWELL: Okay, I guess he's still dining. But anyway, Dr. Homer co-founded the National Initiative for Children's Health Care Quality in 1999. And he currently is President and CEO of that organization. He is Associate Professor in the Department of Society, Human Development, and Health

at Harvard University School of Public Health and Associate Clinical Professor of Pediatrics at the Harvard Medical School.

Dr. Homer, who had been very active in a variety of quality improvement activities, including that at the American Academy of Pediatrics, he's also served on the U.S. Preventive Task Force and a whole variety of activities in this sector. So we welcome Dr. Homer. And he will be an outstanding member of this committee.

Dr. Steven McDonough is here this morning.

Steve, could you stand up? Where are you?

He's here. He must be having breakfast with Dr. Homer.

(Laughter.)

DR. HOWELL: But maybe we could --

FEMALE SPEAKER: They're being sworn in right now. That's why (inaudible).

DR. HOWELL: They're what?

FEMALE SPEAKER: The new members are being sworn in.

DR. HOWELL: The new members are being sworn

in, I'm told, by my consultant to the right. But anyway, as soon as he's sworn in, Steve McDonough will join us. He's a board-certified pediatrician from North Dakota. He has been very active in North Dakota with the Department of Health. And he's served as Medical Director of the Newborn Metabolic Screening Program. So Dr. Steve McDonough will be an outstanding representative from one of those, what I'd call, those large, square states in the middle of the country.

(Laughter.)

DR. HOWELL: And will bring a great deal of information about his activities in the Newborn Screening Committee.

Dieterich Matern is here also. Dieter is Associate Professor of Laboratory Medicine at the Mayo Clinic College of Medicine. He did his genetic fellowship at Duke University. And he is Co-Director of the Biochemical Genetics Laboratory at the Department of Laboratory Medicine at the Mayo. And this committee is extremely familiar with that laboratory, because they have been extraordinarily

active in tandem mass spectroscopy, particularly in reducing false/positives. And he's a close collaborator of Piero Ronaldo. And so, we welcome Dieter. And Dieter is just arrived with his cohorts, et cetera.

Dieter, do you want to stand up?

And that's Dieter.

Steve, would you stand up? We've already introduced you. But you weren't here. Okay.

And Dr. Homer is also here? And Dr. Homer. Okay, fine.

And we have two folks who are here. Cathy Wicklund we're delighted to have here, coming from Northwestern, where she currently heads the Program in Genetic Counseling. Being a pediatrician, I'm always pleased when people start out with a very good career early in life. And that's where Cathy started well, at my old place at the University of Texas in Houston, where she was trained in genetic counseling.

And Cathy's been very active in the field of newborn screening, participating in some Institute of Medicine activities. She also served on the

Secretary's Advisory Committee for Genetics, Health, and Society and currently is very active in the Institute of Medicine Round Table on translating genome-based research and health.

Cathy, would you stand up?

Cathy's sitting here in the front row.

And then, the final new member of the committee is Andrea Williams, who is the Founding Executive Director of the Children of Sickle Cell Foundation, an organization that's committed, obviously, to the well-being of children with sickle cell disease. Andrea has been very active this sector for a long time and currently serves as a member of this group's Education and Training Subcommittee and has been very involved in a variety of issues of newborn screening, with the particular interest and expertise in sickle cell disease.

And, Andrea, where are you? You are here, I know.

There's Andrea. Thank you very much, and so forth.

So that outstanding new group will be

joining the committee. And, apparently, they've been sworn in, which is an excellent sign.

(Laughter.)

DR. HOWELL: Let me also introduce some folks sitting at the table today. Sven Peterson is at the very end, who's the General Counsel from HRSA, representing this sector of HRSA. So we're delighted to have Sven here. And I'm told he'll be here with regularity.

And representing Dr. Wakefield is Sarah Linde-Feucht. And so, we're delighted to have Dr. Wakefield, who is Director of HRSA, having her represented here today.

We have, in addition to the distinguished group coming, we have some longstanding and dedicated, and exemplary members of the committee who will be departing: Rebecca Buckley -- Becky Buckley has been very active in this area; Ned Calonge, Tracy Trotter, Gerry Vockley. And this will also be my last meeting as Chair.

The first order of our business today is to approve the minutes of the May 2011 meeting. And the

committee has had those for some time. And, I believe, you had the chance to look at them.

Can we have a motion to approve them?

MALE SPEAKER: So moved.

DR. HOWELL: Seconded the move?

MALE SPEAKER: Second.

DR. HOWELL: Those favoring, say, "aye."

CHORUS OF VOICES: Aye.

DR. HOWELL: Any abstentions?

(No audible response.)

Any nays?

(No audible response.)

Thank you very much.

We have a lot of committee correspondence that I'd like to spend a little time on. She wants to do housekeeping before we do this. Okay.

(Laughter.)

DR. HOWELL: We do want a neat house.

DR. COPELAND: Yeah, we want a neat house. I'm Sara Copeland. I am the new Executive Secretary. And I will try not to mess this up too badly my first time. So housekeeping notes: when exiting the

general session, the restroom is down the hall and to the left. The Altarum staff will be at the registration desk to direct and assist you and answer any questions. And there's also a get well card for Alaina Harris, who is one of my staff members, who had a stroke back in July. And so, she is recovering remarkably well. But anybody who knows Alaina, knows that she's incredibly social. So she would love to hear from any of you.

Please note we are not able to provide wireless access in here, except for the committee members. Part of the hotel offers complimentary wireless upstairs.

Continental breakfast and lunch will be provided for committee members and presenters only and will be in the Potomac Room Thursday and Friday, just down the hall here.

Subcommittee members, our meetings will be held from 3 to 5 p.m. Labs, Standards, and Procedures will be in City Center 1. Follow-up and Treatment will be in the New Hampshire Ballroom. And Education and Training will be in City Center 2.

If any of the presenters have changed their presentations after submitting them, please saved the revised copy of your presentations on the laptop so we have an updated copy with your name included.

Committee members, organizational reps. and presenters should have received a thumb drive or a link to the briefing book. We do also have a supplement to the briefing book on a thumb drive out front that you can get. If you don't have one or you need to update the supplement, please feel free to go get it. And also, as is always the case, please silence your telephones.

DR. HOWELL: Thank you very much, Sara.

Let me spend a little bit of time with you on the correspondence that we've had. We've had four important correspondence: number one, the Secretary's response regarding screening for sickle cell disease carriers. The second was the Secretary's appreciation for the report we prepared regarding SCID; and, number three, the Secretary's response to our recommendation that HHS coordinate newborn screening emergency preparedness activities as defined in the newborn

screening contingency plan with HHS National Response Network.

And the fourth bit of correspondence, actually, came to me yesterday at a quarter of 5. And that is the Secretary's response to our recommendation concerning critical congenital heart disease and screening for that condition. And I'll spend a little bit of time. We've put the actual copy of the letter at each of the members' desk. And there are other copies floating around for those of you who haven't seen it.

I must confess that I commonly hear that something on YouTube has gone viral. And I must confess I think this letter went viral, because, as I had scarcely gotten the letter from the Secretary, when it started appearing in many forms many places. So it's created a great deal of positive energy. And I think that there are several things I'd like to comment about.

Number one, the Secretary's response to our recommendation is extremely positive. And the first and critical thing is that in the middle of the first

paragraph, she says, I have based -- commenting on the background, and so forth, "I have decided to adopt the committee's recommendation to add critical cyanotic heart disease to the recommended uniform screening panel." So that will be the second addition to the panel that has been made formally.

And, importantly, during the course of our recommendation, there were four additional recommendations for action by the National Institutes of Health, the CDC, and HRSA to address evidence that we identified as necessary, as this implementation goes along. And, quite remarkably and enthusiastically, the Secretary has accepted all of those recommendations and has appended to the letter, that was sent to me that you see, a specific report from the Interagency Coordinating Committee that commented on each of the areas that we recommended, that involving research, surveillance, screening standards, and infrastructure, education and training.

And in each of these, there have been identified organizations within the federal government who has responsibilities to carry out these functions.

And, interestingly enough, I have not seen the Secretary in the past make such a specific recommendation that says that she will instruct these agencies to carry out these tasks. So I think that we are all very excited about this positive response. And we'll look forward to seeing critical cyanotic congenital heart disease get on the panel and be implemented. And I think a number of these areas of interest will be evaluated as that comes along.

Would anyone like to comment about that recommendation? The people around the table have the thing, and it's a very positive recommendation. And we are pleased that the Secretary has been so supportive.

I think the recommendation that was sent downtown was a very strong one. The implementation program that was organized by the committee, with the help of many other professional groups and so forth, really laid out a very nice pathway to look at what needed to be done and how to do it, and so forth.

Jeff?

DR. BOTKIN: Yes, this is wonderful news.

I'm wondering whether, as these other activities are conducted with the different agencies, whether this committee has an ongoing role with evaluating those data as they are generated with the other activities.

DR. HOWELL: I would hope so. But the thing is is I don't know how that's going to be implemented, and so forth. Obviously, the individual groups at CDC and NIH, and so forth, will be organizing these activities, and so forth. And I would -- it would, certainly, make a great deal of sense to coordinate those results through this committee. And I would hope so. But I don't know that there's any formal -- the Secretary recommends that the committee continue to be very involved in this sector. So I would hope that would happen.

In response to the sickle cell carrier recommendation, the Secretary states that she's very pleased to support our first three recommendations. That is that individuals should know their medical risks for various disorders, including the carriers, say, for sickle cell disease. The second was the evaluation and screening for sickle cell disease and

other genetic conditions should take place within the individual's medical home. That was our recommendation. And that would involve counseling regarding the implications of the information for the individual and the assurance of privacy.

And, thirdly, as a part of the individual's annual medical evaluation for participation in sports, all potential athletes should receive education on safe practices proved for the prevention of exercise and heat-induced illnesses. Those were our key recommendations.

She felt that two of our recommendations were not ready. And she recommended that this committee work with the Sickle Cell Disease Association and other relevant health -- HHS agencies, athletic associations, and community-based and health care professional organizations to develop guidelines and educational resources regarding sickle cell trait in all persons and that the National Institutes of Health and the CDC prevention conduct research to ascertain its own athletes with sickle cell trait are at increased risk for exercise-related death. So

those are the two recommendations that she felt was not responsive.

Now, she, however -- her response, she recently unveiled a department-wide initiative to improve care for individuals with sickle cell disease and that this initiative builds on ongoing activity by enhancing coordination and integration of these activities. And she's hopeful that this interagency effort will improve the knowledge base and related health impacts of sickle cell trait and inform future efforts related to our -- two items.

As you recall at the May meeting, the Secretary referred both the residual blood spot as well as the cardiac recommendations I've just discussed to the uniform HHS Interagency Coordinating Committee on Newborn and Child Screening. And that committee, as you know, includes NIH, CDC, HRSA, AHRQ, and FDA. And so, the dried blood spot has been referred to that committee. And we've, obviously, heard back about the heart disease one.

And there are other articles in your book for interest. One is Andrew Ewer's article on, "Pulse

Oximetry Screening for Congenital Heart Disease in Newborns and Infants." Dr. Ewer presented this at the Heart House meeting. But you have a copy of that article, which has now been published. And the other article is, "Strategies for Implementing Screening for Critical Congenital Heart Disease," which has just been published by the American Academy of Pediatrics with Alex Kemper as the senior author.

And we've heard about the housekeeping things, and so forth. And as this is our 25th meeting, we have a considerable history to celebrate and much more to accomplish. And, given that this is our 25th meeting and the great deal of transition, we were planning to have an opportunity to celebrate the past, discuss the present projects, and reflect on future opportunities.

We're going to begin by reviewing the past of newborn screening and the Secretary's Advisory Committee on Hereditary Disease in Newborns and Children. And we're first to hear from Dr. Coleen Boyle from the CDC. And I trust that Coleen is on the phone.

DR. BOYLE: Yes, I'm here. Can you hear me?

DR. HOWELL: Oh, we can hear you well, Coleen.

DR. BOYLE: Oh, wonderful. Wonderful.

DR. HOWELL: We can hear you better than when you're here. You must have a good connection.

(Laughter.)

DR. BOYLE: Well, I'll have to stay away more often, then.

DR. HOWELL: No, no, no. Coleen is going to review the -- list the advances in maternal and infant health as one of the past decade's 10 great public health achievements.

Dr. Boyle?

DR. BOYLE: Oh, wonderful. And, actually, I had one slide. And I don't know if that's projecting.

DR. HOWELL: It is.

DR. BOYLE: Okay, wonderful. And I think this is very appropriate in terms of the introduction that Rod just gave us in terms of highlighting the committee's achievement.

So CDC, as part of its efforts to highlight

achievements in public health, at the end of each decade, identifies those key contributors that have really helped advance public health. And they are in 10 categories. They include things like vaccine-preventable diseases, tobacco control, motor vehicle safety, cardiovascular disease prevention, cancer prevention, emergency preparedness, which is really a new category in this decade, and then, maternal and child health.

So as part of the efforts to highlight what we actually achieved over the last decade, 2001 to 2010, we did highlight -- and this is in collaboration with our other agencies and reaching out to them. We highlighted, really, the achievements that this committee helped move forward. And that was in terms of improvements in technology and the endorsement of a uniform newborn screening panel for diseases that has really led to earlier life-saving treatment and intervention.

And we estimated that about 3,400 children are identified each year on, again, uniformly across states with selected endocrine and genetic disorders,

that the panel itself established the recommended uniform panel as of April 2011. All states and territories were screening for 26 disorders across those states.

And then, we also highlighted, over the decade, the achievements made in progression of screening for a functional disorder -- and that is hearing loss -- from about 47 percent at the beginning of that decade to 96 percent and also acknowledging that the follow-up aspects have also increased over time from about 52 percent in 1999 to 69, close to 70 percent in 2008.

So, again, I think we're, clearly, moving in the right direction with that. So I think that just is a nice way to reflect that the work of the committee and the work preceding the committee have really helped to standardize newborn screening for the United States.

DR. HOWELL: Coleen, thank you very much.

Are there any questions of Coleen about this commentary from the CDC? It was very gratifying to see the expansion in newborn screening be identified

as one of the really big public health advances, and so forth. And, again, I think this committee has, certainly, participated in that activity, et cetera.

Any further questions or comments about that?

Coleen, thank you very much.

DR. BOYLE: Oh, you're welcome.

DR. HOWELL: We're sorry you're not here, but we'll see you next time.

DR. BOYLE: Okay.

DR. HOWELL: Arguably, one of the most important areas that the committee has worked in has been to develop patterns of evidence review for rare conditions. And we're going to move now and hear from a number of folks in that sector. And we're going to hear first from Jim Perrin, who's going to discuss history of the evidence review process and the External Evidence Review Work Group.

DR. PERRIN: Thank you very much, Dr. Howell and committee members. It's nice to be here with you this morning and to talk a bit about the recent history in this area.

So, as a background to what we've been doing in the last four or five years with respect to trying to provide as clear and transparent evidence as possible to help the committee make the very difficult decisions you are faced with with respect to new conditions, in 2007, the Maternal and Child Health Bureau entered into an agreement with our group at the Mass General Hospital for Children, with our collaborators as well at the Duke Clinical Research Institute, to outline and test a process for systematic evidence development, evidence review and evidence development, to help the committee with the best possible evidence to deal with its decisions.

And I do want to acknowledge a few people in the room. Alex Kemper, who'll be speaking after me has been an incredibly helpful partner in this for a long time; Alex Knapp, who has really been our Staff Director and very much keeps many things together in some very useful ways. Ann Comeau, who's been a member of our team from its beginning, is also here. It's been a very interesting group of people working together.

In 2008, after we had, sort of, developed a process and listened to a series of questions and went through those questions with the help of review by this committee, the bureau expanded the scope of our relationship to include our work on developing specific evidence reviews to help inform the Advisory Committee in their decision making. What have been some of the guiding principles from the very beginning of this activity?

One is to adapt, as much as possible, established evidence review processes for screening or treatment programs, recognizing, of course, the special challenges regarding evidence about rare diseases. So much evidence review deals with fairly common diseases, or fairly common processes, where one is likely to have randomized control trials. And that becomes, in many ways, the coin of the realm in trying to make appropriate decisions about what works and doesn't work. And, of course, in the rare diseases that this committee addresses, in general, there are few, if any, randomized trials. And there's a whole different level and way of weighing evidence.

We've also tried to provide for you and for the public, in general, as much transparency as possible in our operations, so that you know exactly what we've done, how we've gone about data abstraction, and the ways that we've approached the review of the data that we've pulled together. And we have invited public access and input into the process in some ways that I'll share in a moment.

Members of the group are listed here along with Ann. Nancy Green has been a partner from the beginning. I should have commented on Lisa Prosser, who is also here today, who's really brought a real attention to some of the issues in costs of screening, for which we have usually very limited evidence -- Denise Queally, who's been a consumer representative on our team; and Danielle Metterville, who's a genetic counselor, who's also been a member of our team.

The objectives of the reviews that we have done have been pretty clear. We want to provide timely information to you folks in your consideration of additions to routine newborn screening. We've had a very clear conflict of interest policy, in some

ways, modeled after what the Institute of Medicine has required for committee membership for any of their evidence committees.

And the conflict of interest, which some people have not been very happy to get those forms from us -- but the conflict of interest has included all of us on the staff, for sure, anyone whom we have addressed as consultants to our project -- we have an external consultant group for us -- and, importantly, anyone else we've talked to about the particular condition, because many people in the consumer community, or many people in the investigator community, may, indeed, have conflicts. And we have tried to be aware of those and to bring those to our table in consideration of the evidence that we obtain.

And I think it's very important to understand that where we have asked for information from outside investigators, for example, we've not asked them to review the kinds of summaries we have provided of the evidence. That's really for you folks to do. We have asked them to check the accuracy of the facts that we report as evidence.

So, again, no one external to our group has had the ability to, sort of, influence what the process is, besides providing evidence. And, again, all actual decisions, of course, are made by the Advisory Committee. Our group makes no recommendations. We try to provide you with as transparent data as possible.

So, as we start the process, we have generally worked very hard to define the key questions and to come up with a case definition, which has been easy in certain conditions and extremely difficult in other conditions, to figure out if there really is a well-accepted case definition in the literature, among investigators. And, indeed, we'll talk later on together about ones for which there are real difficulties in case definition.

We have had a case definition group, essentially, bringing in a few experts early in the process. And we try to come up with a case definition that we develop. We bring it back to the Advisory Committee's Nomination and Prioritization Committee so that that team can make sure they agree with how we

have really tried to define, develop a case definition to carry out the reviews.

Our review methods are pretty straightforward. There are, sort of, two pieces to the process: the literature review and then, the discussion with outside experts in the area. And we typically do the literature review first, so that we feel we have a pretty clear understanding of what the known information is in published literature and what are the key questions for which there aren't answers we would like to address without experts. We have generally used measures of these resources, Medline, other citations.

We've typically had a 20-year perspective in most of our work. We have included, really, only peer-reviewed, published literature. We have limited it to English language studies, only ones that involve humans, so no animal model studies.

We have reviewed review consensus statements or proceedings of conferences or other such activities, not as evidence, but rather as guides for some of the key questions in the field. And they

often have additional references that we've used to go back to to make sure that we know whether it's high-quality published evidence.

So pertinent material that we use must meet our case definition and must address some of the key questions we've defined. Our abstraction method is pretty straightforward. Three investigators review all abstracts and independently abstract a sub-set of approximately 20 percent of all articles. And we use standard quality assessment methods, which we had described in the past to this committee.

We then have, typically, contact with experts outside the systematic literature review. And these are basically key investigators, people who have published extensively in this area, are working with populations with these conditions, who have done screening. This is not limited to U.S., so we've had conversations with people in Europe, Japan, and elsewhere, if the condition particularly relates -- if their work particularly relates to that commission.

We've also worked with advocacy groups to understand what their understanding is of the evidence

in a particular area, what they view to be the key questions, and where they think that there is some evidence to support those key questions. So this is a fairly systematic approach to gathering additional evidence from experts.

And, in general, we've also asked them to provide this, to the degree that they're willing, with raw data from unpublished sources. Now, this, of course, is a tricky problem, because most investigators don't want to share unpublished data before they've gone ahead and published them. And if we actually use the data and present the data to the A.C., it becomes part of public record. And, therefore, you can understand how delicate the balance is on our ability to get raw data.

We've really sought it actively where we've felt that raw data would help us provide better evidence to this committee about what's happening with unfollowed populations or children who aren't being treated, things like that, which can be extremely valuable for this committee's understanding. We try to get that. And that's probably been our highest

focus.

Our evidence review results and summary have tended to follow this presenting the results, again, in the ordering content of the main questions that we've agreed upon with you. The decision analyses and decision model findings, outcome tables, and summary, then, with key findings, which we're now trying to present to you in summary and table form, and to indicate where evidence is absent, where there are often many gaps in evidence for many of these conditions, and what information would be most critical, what we don't know and what we do know and what's the level of uncertainty and what new information, what new studies would most help committee decisions.

We don't tend to say to you, "Golly, there's a lot of absent evidence here, and more research is needed." We try to say, more specifically, "We think that the research that's particularly lacking is this, and these are the studies that ought to be done." Again, all decisions are made by you folks. We make no decisions. We make no specific recommendations as

to what the A.C. should do.

So what are the evidence key questions? The over-arching question, of course, is, is there direct evidence, direct evidence that screening at birth leads to improved outcomes for the infant or child screened or for the child's family. That's predominantly the question that we've addressed in all of our reviews. The questions relating to the specific condition, is, again, is there a case definition; what is known about the natural history and spectrum of disease, with and without treatment; what is known about the incidents and severity of the health impact of the condition.

With respect to the screening test itself, we typically will look at the analytic validity of the test, the utilities of the test, and sensitivity specificity, predictive values, the clinical validity of the screening test by itself, and then, in combination with a diagnostic test, the timing of screening, when is it best done, and why is it best done at that particular time, what is known about follow-up. And we tried to identify for the committee

if there be population-based screening evidence rather than clinically-based or other selected population-based screening evidence. And this has been critical for a couple of the conditions that we've addressed.

With respect to treatment, we've looked at the question of does the treatment of screened, detected condition improve important health outcomes compared with waiting until clinical detection. And that's relevant for things like SCID, for example.

Are treatments standardized and widely available, and, if appropriate, FDA-approved? And a third area, which has been a real challenge, but very interesting, is are there sub-sets of affected children more likely to benefit for treatment who can be identified through testing or clinical findings. And then, we've tried to understand more about benefits, harms, and costs. What are the benefits of treatment? And this, in many ways, reflects the maximum number of potential beneficiaries.

What are the harms or risks of screening, diagnosis, and treatment? And what are the costs of any of these elements? And, again, repeating what I

said before, these are areas for which the harms and risks we often have very, very limited information, and even less for costs.

So what are the real challenges that we faced? One is the lack of a very clear case definition. So Krabbe Disease is a good example of one here, where there's a very wide variation across a spectrum of disease severity for people who are screened positive for Krabbe Disease.

Second is that these conditions are extremely rare, in those cases. And they often -- almost all that we've identified have high severity. We're not really examining, at least to this point, low severity conditions. Many of them have fatal outcomes. So there isn't much debate about whether these are important, clinically, from the viewpoint of children or families who are affected by these conditions. But as rare conditions, again, there's a lack of randomized trial in almost all the cases that we've worked on.

A third issue is, really, the lack of decent population studies of screening for rare conditions.

And to do them right, it often requires several years of data, even in large states, to document the sensitivity and specificity. And this, in fact, was one of the issues in the committee's deliberations about whether or not to add SCID to the uniform panel. Indeed, there were population studies, after our original report, that helped to provide better evidence for the committee.

As I said before, costs and benefits are rarely well-documented. And it's also true that, in some cases, Pompe's Disease, which this committee debated in great detail, critical sources of information may be unpublished and very, very difficult to ferret out. We've tried, again, in that case, in particular, to provide you the best possible evidence.

So these are, then, some of the reports that we've done for the committee in November of 2008: Pompe's Disease, severe combined immunodeficiency, Krabbe Disease, Hemoglobin H Disease, critical congenital cyanotic heart disease, which Dr. Howell and the Secretary have commented on this morning. And

then, we are in the midst of finalizing a report for you with respect to neonatal hyperbilirubinemia, which is a challenging evidence review as well.

Briefly, other activities that we've carried out with our group related to this -- one is a publication in 2010 just describing the process that the Evidence Review Group put together for the purposes of this committee, a publication on SCID in Pediatrics, a publication on Krabbe Disease in Genetics and Medicine. We developed a work group back in March, with help from the bureau, to really look again intensively at our evidence evaluation methods. That work group is continuing in certain ways. And then, we had a publication in the Journal of Peds relatively recently on the review of Hemoglobin H.

That's the end of my comments. I just want to say how grateful we are for the opportunity to have worked with the committee. It's been a wonderfully interesting few years. We've learned a tremendous amount from this experience with you. And it's been a real pleasure working with the committee. Thank you.

DR. HOWELL: Thank you very much, Jim.

Are there questions of Dr. Perrin?

I have a couple. One is that you listed a lot of challenges. Which is the most perplexing challenge that you really feel that you still have not made major inroads into approaching?

DR. PERRIN: So I think probably one of the hardest ones and one we're working on actively -- and, I think, Alex will talk about this shortly -- is the weighing of the evidence. So in traditional evidence review terms, the evidence that we have in most cases varies from weak to awful. And so, that's not a satisfactory statement, I think, from the viewpoint of public policy with respect to trying to make some very, very difficult decisions here.

So a real task is to come up with a much more satisfactory way of presenting the evidence to you in a way that clarifies where the evidence may be particularly helpful to you and where the evidence, frankly, is highly suspect. That's probably, from my viewpoint, the biggest problem.

DR. HOWELL: Another more general question -- and that is that this is, as far as I'm aware, the

first, really, big effort to try to look at evidence in rare conditions so people can make decisions. And so, it's a new area. How are your efforts viewed by the hard-nosed evidence review world? What do they think of what you've done?

DR. PERRIN: It's very light and softly. No. Alex Kemper probably can provide a better sense of that, because he's a little bit more tied into some of those groups than I am. But I think we have developed some real credibility for this process within the community. I think that's been very helpful. I think there's a recognition that the work that we've done is, indeed, a responsible, transparent, and tries to make the best use of available evidence. And if Ned has other views on this --

DR. HOWELL: Ned, would you comment? You're a pillar of that community, of this community.

DR. CALONGE: (Inaudible) try to not be hard-nosed. But other than that, no, I think there are about three comments I would make. One is that the rest of the evidence synthesis and translation

community recognizes this is a difficult problem and are thrilled that someone other than they are willing to take it on.

(Laughter.)

DR. CALONGE: The second thing is I cannot understate the value of bringing the kind of evidence that that same group together to discuss methods, as we did last year. You know, Alex can argue that we made only a little bit of progress. But we did make progress. But the most important thing was putting that group in the room to understand the problem and to understand the directions that the group was trying to work on, moving forward, to address the issues of translating and synthesizing evidence in the face of no evidence, but great need.

And so, I cannot underscore -- although he had been nice to come out with this huge, new transformative approach to rare condition evidence. Just getting people in the room to all agree and identify the problem and then providing a launching point for decision making, modeling, and other strategies going forward was key.

So the last thing I'd say is that the group is ongoing. And this ongoing commitment to refining, testing, demonstrating, and evaluating methods in this area is a long-term commitment that, quite honestly, the evidence-based world and the world of rare conditions needs to be wise and make good decisions.

DR. HOWELL: Alan?

DR. FLEISCHMAN: I think one of the great contributions of the Chair and the Chair of our very special Evidence-Based Work Group has been to give credibility to the process that's around this table. Prior to that very structured, very competent, very thoughtful review, there were critics, both in the evidence-based world, but also in the bioethics community, who were questioning the process, I think, inappropriately. But they were still questioning the process.

And Jim's team has brought credibility to a public health problem that needed to be addressed, whether it was going to be done well or not well. And it was done extraordinarily well. And I think we are in his debt and in the Chair's debt for having created

this process that we can be proud of and that the public can respect. When smart people try to make hard choices, they have the best possible evidence. And they still have to make hard choices.

DR. HOWELL: Gerry?

DR. VOCKLEY: One of the big, remaining challenges in the evidence-based process, I think, rests with the individuals that are out in the field dealing with these patients and the families and patients themselves. You know, I'm delighted to hear that we've made some progress within the evidence-based world. But if we can't translate that into an understanding at the level of the real world that says, we appreciate the need.

We are very understanding about the way individuals and groups would like to have their agendas moved forward as quickly as possible, but then, to also have the recognition that, without the evidence, you just can't move forward. And the recognition that this group really does try very hard to move those kinds of agendas forward as best we can -- I hope that both of those, you know, the evidence-

based world and the real world, are moving forward at the same time.

DR. HOWELL: Jeff?

DR. BOTKIN: I guess as time goes on, I've become more sensitive to, sort of, some of the circular challenges that this whole field presents. In other words, trying to make a decision about when population screening is justified, but yet, one doesn't have the data without conducting population screening.

And so, it seems one of the challenges for us has to be, as reflected, I think, in the congenital cyanotic heart situation, which is once we reach some threshold to say it's justified to move forward, to continue to collect those data on that initial implementation and come back and revisit the question once those data are in-hand and think about the possibilities of changing our mind later, at least making that conceivable to say, preliminary data was adequate to initiate those screening programs. We've collected the data. And, in fact, now we can make a more informed decision about whether this ought to be

part of an ongoing uniform panel.

DR. HOWELL: I couldn't agree more with that. And again, I think that a great example of doing this happened with SCID, where it was clear that it seemed to be a very good idea. But a large population study was done that demonstrated, really, a most effective screening test. And that was done under an investigative fashion. And I think the same thing must happen in congenital heart disease so that we have data coming back, and so forth.

But I think that, Jim, your group has just been remarkable, because I think focusing aggressively on getting the best information that's available -- because if you don't make a decision about a serious problem that's ongoing, that's not a good thing to do, regardless. You need the best information to let you make a sensible decision. And I think that's what we tried to do. And I think your group has really done a very good job in doing that.

MALE SPEAKER: It does raise the question again that was raised earlier about the committee's role and purpose in examining new data as they become

available and wanting to stay involved with that process as part of understanding the role of the committee. I think that's -- I mean, we're never going to have enough evidence. That's very clear. And if the committee makes a decision one way or the other, it may help the committee to be able to revisit that as new evidence develops.

DR. HOWELL: I think that everybody around this table is very familiar with the fact that the establishment of the Newborn Screening Translational Research Network was done with this in mind. In other words, that there would be a systematic evaluation of new technologies and treatments, and so forth, in a scientific way that would inform the committee and the country, and so forth. And hopefully, I know that's moving along with a lot of good things, and hopefully, will be re-upped fairly soon.

Becky?

DR. BUCKLEY: Well, I hope that your committee is going to continue with ongoing its work.

DR. PERRIN: We certainly hope so.

DR. BUCKLEY: And your presentation sounded

somewhat final, but I hope it continues. And the reason I ask is that, you know, all the other conditions that haven't undergone evidence review -- I think that, considering his remarks, I think that they should apply to all of the conditions that we're currently screening for.

Having been in touch with a number of state newborn screening people over the past few months trying to get them to establish SCID in their state, I keep hearing from the newborn screeners that so many of the things they screen for -- and they don't ever find very many. And I wonder if there's any plan for your committee to go back and look at some of those.

DR. PERRIN: So the committee will continue. We're looking at some changes in personnel, but the committee will continue, assuming that the Advisory Committee wants it to do so. I think that, as you remember, we, on the Evidence Review Group, respond to the committee's nominations. So nominations can come in from any part of the field. Any type of person, or group, can make a nomination. And it's reviewed by the Advisory Committee's Subcommittee, comes to this

committee for further consideration. And if you believe we should review it, we do so. We don't choose the topics.

DR. HOWELL: There are a number of things out there that should come to the committee soon. And hopefully, the folks in that sector will see that happen, because there are a number of conditions that are going to be on the agenda quickly.

Chris?

DR. KUS: (Off-mike) cost/benefit part, because that's the part which has -- given what's happening today, that's a big issue. And is there -- as we add new conditions and we're, hopefully, improving long-term follow-up, is there a way to get a handle about conditions that are improved and cost/benefit? Or any talks on that?

DR. PERRIN: So I think two or three thoughts. And I might ask Lisa, if that's all right, to respond as well to that question. So, again, remember, our job is primarily to look at evidence, where it exists, to -- we don't have much ability to generate new evidence in our group. So the questions

you're asking are very difficult for us to respond to, because there is almost never any serious published evidence in this area. A couple of exceptions to that rule, but not very many.

It does seem it's a tremendously important question for the committee, though. And it may behoove the committee to explore other strategies for coming up with estimates in that area, because it's not going to be based on published or easily available evidence.

May I ask Lisa Prosser --

DR. HOWELL: By all means.

DR. PERRIN: If you have any additional comments on this?

DR. HOWELL: Lisa has, as, obviously, Jim has pointed out, has been a pillar of this committee along.

DR. PROSSER: Thanks. So tomorrow I'll be talking about how we're planning to move forward in terms of incorporating decision modeling into the evidence review process, so moving beyond just reviewing evidence, but synthesizing that evidence to

provide some additional information to the committee. And, as part of that process, we can talk about where cost effectiveness and generating that kind of evidence would fit into that. But that would, certainly, move the process forward, even one more step beyond where we're planning to go now.

DR. BAILEY: So I would echo the compliments from the committee in terms of the fine work that your group has done. And also, I recognize that you've, you know, published a number of articles about the review process and how you've gone about it, which have been excellent.

I wonder if another product might be, kind of, stepping back from across the different conditions and making some recommendations for either advocacy groups or clinicians or other researchers who have their favorite condition that they would like ultimately to be nominated. And what would be some examples of creative ways that people have gone about approaching rare diseases and studying them and bringing the evidence forward that's been most useful to your committee? I don't know if that would be

something that your group could take on. But I would think that the community would be very appreciative of that.

DR. PERRIN: That's a really wonderful idea. So one thing we are working on is, sort of, a manual of procedures to really take partly the advice we got from the committee that Ned helped us put together and to really try to be more explicit about what we do here. I don't think it'll be user-friendly, frankly, in the sense of being valuable to very many people outside this group in the field.

But I'm just, sort of, wondering whether one could develop a couple of, sort of, public modules of that, one for families and one for clinician/investigators or clinicians. It's, certainly, worth putting on the table. I think there's some real value to that.

DR. HOWELL: I think that's a very good idea, because I think commonly, folks would like -- some folks will approach you wanting to screen for something that, clearly, has some real issues with screening for it. And to outline what you really

need, and so forth, could really be very helpful.

Any more comments for Jim?

Denise?

DR. DOUGHERTY: Yeah, I'm just wondering if (inaudible) Don's comment, about maybe the committee could consider becoming more proactive in, sort of, recommending some research infrastructure or general research protocols that can be used in this area, so that we're not always playing catch-up. It's always a frustration -- it is, if the U.S. Preventive Services Task Force, you know, comes up with a recommendation, says insufficient evidence. But then, there's no translational piece that says, you know, somebody (inaudible) uptake getting that evidence in place before you have to revisit that condition again. So just making some recommendations about how we can get better evidence.

DR. HOWELL: Sharon and then Ned?

MS. TERRY: Also building on Don's comments -- so at the beginning of the process, Genetic Alliance was written into it as a technical assistance to these advocacy organizations to help walk them

through the parts that they're involved in. And we have done that. But we probably could do that in some more visible way or proactive way along the same lines as being proactive rather than just responsive.

DR. HOWELL: And, Ned?

DR. CALONGE: Jim, I actually think this group is likely to lead -- or at least have the opportunity to lead the way of the use of modeling in presenting the groups like this, recommendation groups, with the data from modeling used to make decision making. And people just need to know that, while that's happened a little bit, we're really on the cusp of that. It's not widely accepted. When you do it, you get criticized. And yet, I think it's just going to be an important part of this committee's work, moving forward.

And so, what I'm trying to do is touch all these points together. So you can model anything; right? The only issue is what are the assumptions you have to make. And we're often making assumptions based on only a couple of data points. One is that the condition exists, and, two, that we have some kind

of numerator data that came from somewhere that's usually heavily filtered, biased, nuanced in the ways that don't reflect the, kind of, underlying problem that we might be facing.

And so, I think it's just important to recognize that, yeah, we could put out there recommendations for doing research that would fill in the evidence gaps. But we need to think more broadly. What kind of research would benefit us in terms of the assumptions that we could make better assumptions in our modeling data, which I think we're going to be stuck with for a long time? And so, there's a broader set of recommendations we could put out.

The other thing is it's -- you know, we're always -- what we're trying to do is decrease our risk of being wrong. Okay? And when we say, okay, we're going to add it to the list, the tipping point for us is that we're relatively certain that we're not wrong. And that's okay. So recognize that we're in shades of grey, but we're trying to sharpen the shades so that they're darker or lighter. And that's okay.

And the last thing I would say is, as we do

modeling, it should always be done with the assumption, Denise, that we're going to fill in the data gaps as we roll out something. And I think that was a landmark part of the congenital heart disease recommendation is that we're going to see what we're doing.

And this group's going to have the discipline that, if after we collect data for 10 years and it's a completely different group of people and it's become acculturated in newborn screening and we find out that it doesn't work, which you might, right, because it's always a risk of being wrong, that you're willing to stand out there and say, we're not going to do it anymore. So when you think about the methods, data creation, and trying to be proactive, recognize that it's not going to look like the usual RCT evidence-based world. And it doesn't need to.

But it doesn't mean that we can't continue to be very strategic, evidence-based, and make good decisions that have a great chance of improving health and not just going the other way. Thank you.

DR. HOWELL: Thank you very much.

Jim, thank you for your committee helping reduce our chances of being wrong.

(Laughter.)

DR. HOWELL: Thank you very much.

We are now going to hear from Alex Kemper, who's going to address the history of the other work of the Secretary's Advisory Committee on Hereditary Disorders in Newborns and Children. Alex is at Duke, as many of you know, while he's getting there, there's even a little view there of Mr. Duke.

DR. KEMPER: So good morning, everyone. First, before I get started, I'd just like to recognize that the work of the Advisory Committee has really led to improvements in the lives of children and their families. The Advisory Committee itself has been just incredibly productive. And, in this talk, I'm going to be talking about the other work of the committee.

So we're, you know, now for something totally different, I'm going to get away from evidence. And I'm going to be talking about the work that the Advisory Committee has done. And I should

say, too, that it's really been a privilege of mine to be involved in some of this other work. I, kind of, feel like I'm the groupie for the Advisory Committee. I'm obviously not a member of the Advisory Committee, but I've been involved in a lot of activities. And it's really been a pleasure to see how everything evolves.

So, by way of background, the Advisory Committee has really addressed broad issues related to improving health outcomes through newborn screening. And a lot of that work is done through its active subcommittees, which have developed all sorts of work, including surveys and white papers and recommendations to the Secretary. These subcommittees make recommendations to the Advisory Committee as a whole. And some of these recommendations to the Advisory Committee as a whole then move up to the Secretary.

And so, it just wouldn't be possible for me in the next little bit to summarize all of the other work that's being done through the subcommittees. And just necessarily, I would end up leaving out important things. And so, after getting wise counsel from Dr.

Copeland, the focus of this talk is really going to be on those recommendations that have bubbled up through the subcommittees and have gone to the Secretary.

And, I think, it's also instructive to step back and think about what the purview is of the Advisory Committee and how it developed, especially as new members come on. So the Advisory Committee itself was chartered in 2003 with a broad range of duties. This is from the actual document itself. It's like looking at the Constitution going through these old documents.

But the Advisory Committee shall provide advice and recommendations to the Secretary concerning grants and projects, provide technical information to the Secretary for the development of policies and priorities for the administration of these newborn screening-related grants, and finally, to provide such recommendations, advice, or information as may be necessary to enhance, expand, or improve the ability of the Secretary to reduce the mortality and morbidity from heritable disorders. So that's really quite a broad scope of potential activities. And I think the

Advisory Committee has really (inaudible) been there to do so.

Now, in offense, some of these activities were further defined, but also expanded through the Newborn Screening Saves Lives Act. Sort of interesting historical note: The Newborn Screening Saves Lives Act went through in 2008. But the short title is Newborn Screening Saves Lives Act of 2007. And as I was doing this search on (inaudible) on the act, sometimes I find it referred to as the Act for 2007. And sometimes it's the Act of 2008. But near as I can tell, they're all the same thing.

So the Newborn Screening Saves Lives Act outlines a really broad range of activities, including making systematic evidence-based and peer-reviewed recommendations -- obviously, that's what I've spent most of my time working -- to develop a model of (inaudible) matrix for newborn screening expansion, including an evaluation of the public health impact of expansion; to consider ways to ensure that all states obtain the capacity for screening, short and long-term follow-up; to standardize language and terminology

used by state newborn screening programs; quality assurance oversight and evaluation to participate in developing education, not only for providers, but for everybody involved in the newborn screening system, including families; assessments of costs and effectiveness -- going back to some of the comments that Dr. Prosser was making before -- and coordination of surveillance activities.

So that's a whole lot of activities. And I really think the Advisory Committee has done an incredible job of addressing many of these. So I'm going to be talking about some issues, including health reform and coverage for medical food, education, long-term follow-up, the national contingency plan and sickle cell disease, indeed, making a smattering of other comments as I go through.

And hopefully, at the end of this, it would be very interesting for me to hear from the rest of you about activities that you think that the Advisory Committee has been involved with that have really made a big difference. Because, like I said, just by necessity, not everything is going to be included.

So the issue of health reform and coverage for medical foods has been challenging. The first letter that I found particularly addressing this was from May of 2009, where it states that the Advisory Committee desires a more uniform approach towards coverage by health care payers of medical foods and foods for those conditions recommended by the committee and specific amendments to Medicaid legislation to ensure more uniform coverage by state Medicaid programs.

In response from the Secretary in October 2009, there was a letter that basically said -- I'll read it here. "It is understood that the committee feels that policies are needed to address gaps in coverage for items that are a vital component of medical management, but not typically included is medical services for the disorders identified through newborn screening." And then, skipping to the last sentence, "However, the committee's recommendation to enact legislation go beyond the department's authority. Therefore, I am neither adopting nor rejecting the committee's recommendation."

So, although there is general support, it just wasn't, at that time, within the purview of the Secretary to do so. Now, of course, a lot of things have happened since then, including the Affordable Care Act, which I will get to in a second.

So in March of 2010, there was a follow-up letter to the Secretary from the Advisory Committee addressing these things, which included encouraging CMS to convene an expert panel to examine coding challenges around newborn screening and to standardize health information exchange. The second one was to encourage CMS to develop and pilot a payment method for integrated systems of care coordination through the medical home framework for children diagnosed with heritable and congenital disorders as a result of newborn screening, to encourage the adoption and further definition of the newborn screening use case. And this was part of expanding the health information exchange and meaningful use around newborn screening.

And finally, here again is the medical foods issue -- to support, if allowable, the closure of gaps in insurance coverage for medical foods and foods

modified to be low in protein, as recommended by the committee back in April. In response, the first three recommendations were accepted.

And within that letter, there was a particular note that the lack of coding in billing, clear guidance was an administrative burden, that the medical home models within the letter were specifically highlighted as something important. And it was clear that the benefit of electronic exchange of data was seen as a way to improve care for a nation.

But what about medical foods? That's been an important issue to the Advisory Committee. So in response to the medical foods issue, again, the recommendation was not accepted. It was understood that there was a policy needed to cover the gaps.

But all this needed to be enacted within the context of the Affordable Care Act. And the Secretary stated that my forthcoming response to the June 14th letter will address this further and that CMS would be asked to review state Medicaid programs to determine if there's an opportunity to improve federal guidance

around this area.

And in, again, another letter to the Secretary emphasizing this, medical foods issues and the importance of it, the Advisory Committee wrote that the committee believes that our nation has a special responsibility to assure evidence-based treatment for individuals identified with these disorders and emphasize the need to provide these life-saving treatments over the lifespan of the individual.

And, in response, again, the information was deemed to be helpful. And the Secretary understood these issues. But still, there's a process that needs to go through. And serious consideration is being given to the issues raised.

So, you know, I think this illustrates that this is a complicated process, especially around providing coverage for medical foods, which is vitally important to many of the individuals that we identified through newborn screening. One of the great things about the Advisory Committee, though, is, beyond just making these recommendations to the

Secretary, is its relationship with the regional collaboratives, which are funded by HRSA, to improve the process of newborn screening.

The regional collaboratives -- and I'd specifically like to point out the work of Dr. Sue Barry and Dr. Ronny Singh -- have done a lot of work to collect barriers and understand what is challenging families around the receipt of medical foods. And then, as a result of that activities, they've developed individual projects within the regional collaboratives to help families. And then, all this is tied back through the National Coordinating Center. And, maybe if we're done, Dr. Rotchin can talk a little bit about that -- as a way to disseminate best practices to the other regional collaboratives.

So I think that the Advisory Committee is making -- through these recommendations, having a very important and profound effect through the regional collaboratives. And I think this is a good example to illustrate how the Advisory Committee works with the subcommittees.

So, for example, the Long-Term Follow-Up

Subcommittee -- I'm, like, getting its name wrong, I'm sure -- has defined long-term follow-up as including care coordination through a medical home, evidence-based treatment, the use of continuous quality improvement, and new knowledge discovery. This was a really important step by the Advisory Committee, because it really laid out the issue that newborn screening isn't just case identification, but making sure that children, through their lifespan, get the best care that they can get.

And by defining long-term follow-up, that's really helped the regional collaboratives in their activities and has facilitated partnership. And, for those who don't know much about the regional collaboratives, I did just put up a map here of them.

In terms of education, I think it's interesting that the early work of the Advisory Committee really anticipated the Newborn Screening Saves Lives Act. I have a sample of the letter from December of 2006, where there is an emphasis on developing and funding a mechanism to study the distribution of existing newborn screening educational

material and the acquisition of knowledge about newborn screening by expectant parents in the context of the health care provider/patient relationship.

And I think that that's been a very important theme that's run through the work that the Advisory Committee has done and, certainly, been the focus of some really great work that Dr. Terry has done. And if she wants to talk about that later, that would be excellent as well. And I know that there's going to be a longer session as well.

The Education and Training Subcommittee also developed a report describing the need for primary care education that was endorsed by the Advisory Committee. And that led to funding through HRSA of the Genetics and Primary Care Training Institute. I believe the American Academy of Pediatrics, is that right, has won that grant?

And, again, this illustrates how things can bubble up through the subcommittees, and then, after recommendation by the Advisory Committee, can lead to a funding of new endeavors. And hopefully, there'll still be dollars out there to continue that kind of

work.

I'll briefly touch on the national contingency plan that was presented to the Secretary in August of 2010, which recommended that each state have the newborn screening contingency plan. Of course, I think there was a lot of thought about this that developed on the heels of Hurricane Katrina. One of the key things there is that the CDC will, with support from HRSA, will lead efforts to coordinate implementation with the assistant secretary for preparedness and response.

The regional collaboratives themselves have taken an active role in disaster planning. And I know that there have been a lot of these tabletop exercises, where they simulate a disaster, and then, feedback within the regional collaboratives can develop systems in case of a disaster.

Now, let me see if I can go back. Yeah. There was a letter, which I didn't have time to add in, that just came back this month, where the Secretary essentially further endorsed the contingency plan.

Dr. Howell spoke just a little bit ago about the sickle cell trait issue. And so, I won't go through the letters again. But I think that this is another example where the Advisory Committee took on a very complex issue, that is testing athletes for sickle cell trait and came up with very common-sensical recommendations, which are now, by and large, being adopted by the Secretary.

There has been so much work around dried blood spots that I'm almost hesitant to talk about it, especially with such (inaudible) with Dr. Botkin here. I would just embarrass myself, I think. But the Advisory Committee has recommended that the states develop policies related to access of dried blood spots (inaudible) physician, education health care providers and families, documentations of parents' wishes, and has recommended that there should be a national dialogue.

Again, Dr. Botkin, you talked a lot about this -- and explore the utility and feasibility of a voluntary national repository.

In April, there was a letter from the

Secretary to the Advisory Committee that said that those particular recommendations weren't ready for adoption, but things were referred to interagency coordinating committee. But I think it's important to emphasize that the work of the Advisory Committee, again, has really helped the regional collaboratives and the National Coordinating Center in thinking about these issues. Certainly, the National Newborn Screening and Translational Research Network has also been addressed by many of the subcommittees of the Advisory Committee and projects funded by the Health and Human Services, including the meeting that Dr. Botkin just held in the great state of Utah just this past week.

So, you know, again, I'm, sort of, sheepish, because there's so much stuff that the Advisory Committee has done. And there's no way, within a short period of time, that I can highlight all of them. But what I do want to say is that the Advisory Committee and its subcommittees have been incredibly active and productive. I do believe that the work has led to improvements in the care that children and

their families receive.

I think that there's still a lot of important areas to address. I think that, for example, the medical foods issue is not going to go away anytime soon, but that there is a lot of opportunity for thinking about coverage for these life-saving therapies.

I do think also that there is this good model of success that's developed, that under guidance from the Advisory Committee, the subcommittees have developed these reports and that these either go, if they're approved by the Advisory Committee, to the Secretary, who can then act on it. But there's these other venues where lots of activity goes through the regional collaboratives and the National Coordinating Centers, which really look to the Advisory Committee to, kind of, blaze a path through. So -- oops, I'll do this back up.

So, I guess, at this point, I'd like to just stop and see if other people would like to chime in on, you know, this, sort of, other important work and if there's something that should be highlighted,

especially for the new members as they come in to the committee.

Dr. Howell?

DR. HOWELL: Thank you very much, Alex.

Comments or questions of Alex about these reports?

I think you must have said it all. Thank you, Alex. I think you've been a very tried and true groupie. And so, we hope that you'll continue.

(Laughter.)

DR. KEMPER: I feel like -- it's like when (inaudible) said, "I remember all the other ones."

DR. HOWELL: Yeah, that's right.

DR. KEMPER: And I look forward with great anticipation.

(Laughter.)

DR. HOWELL: And hopefully, you can even get more groupies to join you. Great.

(Laughter.)

Ned?

DR. CALONGE: If I could make a (inaudible), not to Alex, but to the group, especially the new

members, as we are facing this pivot. So this would be a great time to actually examine the subcommittees, their scope, and their work and determine if these are the right subcommittees, whether or not there's additions or changes to the charges. And I think anytime there's this big change in membership, it's the perfect time to do that.

So saying, I would pitch the issue that every other group that does recommendations I've ever been on has a Methods Subcommittee. And if you put the last two talks together, that would be something I would hope the next Advisory Committee might think about adding, so those of us who aren't laboratorians, but are assigned to laboratory standards, would have someplace to go in the afternoon.

(Laughter.)

DR. HOWELL: I thank you very much, Ned.

Any other comments to Ned's comment?

We're now going to hear from Jana Monaco. And Jana is going to talk about the role of engaging parents and consumers to weigh in and acknowledge viewpoints. And Jana is, of course, a former and very

active member of this organization.

Jana, good morning.

MS. MONACO: Hopefully, I'll get this right. Hi. It's great to be here and sit at the table again one more time with everyone. I was asked to come and speak on the consumer perspective because I take great pride in having attended all the meetings except for one last January, which was for good reason, when my son was having surgery. But being part of these meetings for the past seven years has enabled me to really see and appreciate the growth in where the work of the committee has gone. And, I think, all the evidence that has been presented over the years has really spoken for itself, and the achievements and where we've come in newborn screening. So I'm just going to give you just a little bit of a perspective, from a consumer's end of things, of where, I think, we've been and where this committee is today and, hopefully, where it will go. Hopefully, I remember how to do this.

I decided to take Tracy's view on things and put a little spin on things, after working with him.

I never really liked (inaudible) definition of a consumer. I really didn't see myself as a consumer when I first came into this, when I sat at this table seven years ago sharing the story of our traumatic experience with my son, Steven, who was undetected at birth and experienced a severe metabolic acidosis at age three and-a-half. And it was just 10 years ago this year that we brought him home.

And then, we had our daughter, who we did seek screening when we were expecting her. So we have two different perspectives. But I'm still being identified as a consumer. I've come to adopt it and appreciate it over time. But I wanted to give you a definition of a consumer. And I wrote it twice at first when I looked it up.

And it was one that -- one acquires goods or services to for direct use of ownership rather than for resale or use in production manufacturing. And I emphasize it a second time, because thinking medically, which the definition is -- or, in the medical perspective, a patient or person who requires medical assistance. When you think of newborn

screening, they are people, we are people seeking some sort of service for direct use or ownership. And that is to save our children's lives. And that's what it's about.

From the committee perspective, it's members of the public having a special expertise about or concern with heritable disorders. So most people coming to the table as a consumer have a very distinct kind of expertise. And most aren't very good (inaudible) this committee.

When you think of consumer advocate of newborn screening, they take on various roles and various definitions. They are patients and families. And we definitely consider ourselves the experts. And I think most people in the field have definitely commended us and given us that title of being the experts on these diseases in our children.

Some consumers are the parents, like myself, who have children with physical and neurological complications due to lack of screening, severe and not so severe. And they're also parents of deceased children who were not screened and either died at a

very young age after birth or even a little bit later. They're also parents of affected children or parents that were detected early -- I wear that hat, too, but only by the previous traumatic experience.

And then, we also have the adult patients, who are living with undiagnosed disorders or who are being diagnosed as adults, thanks to the progress in the area of inborn error metabolism and heritable disorders. So you see, there are many hats that consumers wear and how we as patients and families come to the table with.

If I were to be a consumer of products or goods on the outside, I would be looking at the consumer reports for different kinds of products. So I thought I would give a little consumer report on the committee from when it began and to today.

So when I think back and I look at the inaugural committee when I first got here, giving my five-minute public comment to, hopefully, it would make a pretty good impact -- along with other family members, the majority of states were not doing expanded screening. It was a trickle effect in some

states. The most weren't -- the supplemental screening information was not provided to families, unless you happened to stumble upon it and were someone very savvy at the Internet.

It was a very high number of diagnosed disorders in the E.R.s and ICUs with children in crisis. And many didn't make it, and most had very negative outcomes.

There was a consumer member on the committee, and the public comment was really the only opportunity for that input. And so, that public comment has been really vital and critical to the consumers, because it was your opportunity to provide your voice to help move this committee along.

When I look at the 25th meeting today issue, there's a lot more to it. The ACMG recommendations to states to provide -- to inform a supplemental screening came after that very first meeting. And that was triumphal to those families of us who were hoping that this committee really was committed to its work.

All states have some sort of expanded

screening now, which we're very excited about. Babies are being diagnosed with newborn screening. They're not all ending up in the E.R.s and ICUs, being detected.

The Newborn Screening Saves Lives Act was implemented and passed (inaudible) then the consumer members on the committee. And then, we have consumers integrated in all three subcommittees of the Advisory Committee, which was really wonderful to start plugging in these voices in the various aspects of the work of the committee.

And the consumer voice has also been included in regional collaboratives throughout the country and committee initiatives like that of the clearinghouse with Genetic Alliance. The medical profession and the public are far more educated on newborn screening in these heritable disorders than ever before. And we can attest that to the great work of this committee.

It's not done, but we definitely don't encounter those kinds of responses that I, myself, encountered. "Oh, you know, those disorders are very

rare. You'll never see them again."

I wanted to put this quote in here, because it takes us into that area, because children that are being screened are actually growing up. And they are reaching adulthood. And this was a recent response that I was given by an adult who was diagnosed with his disorder at a late age. And this man is in his fifties.

And he said, "If you are an adult with an O.A., it's just about impossible to convey an urgency to the medical profession. The local resource would like to see me in seven months, for example, and it's cruel. In most cases, but not all, as your family members with an O.A. become adults" -- in this respect, it could be any disorder" -- the main protection they have, which is you, the parent, will no longer be in the same house."

"The voice of you as a patient will never be as demanding as a parent or a child. The interest in a patient must not just be when they're on a gurney in the E.R. You do not have time to educate the E.R. staff," which really emphasizes the criticalness of

education and training, because this is a reality that many people are still experiencing, especially those adult patients, but even those with children, which we really are starting to address these issues when talking about the medical home.

Looking at advocacy groups, extending beyond just the basic consumer, they are a representation of the diversity of consumers, both pediatric and adults. They come with very disease-specific categories. These groups have specific needs and concerns that are related to newborn screening all the way from, whether it's screening to the follow-up and treatment, the medical foods issues.

There is a critical entity of committee -- they are a critical entity of the committee discussions to help guide and know where are the hot spots that we really need to work on. And they often come with firsthand experience and expertise, because the consumers truly leave this room every day and go home, and they live with these disorders. And they live the life.

To increase consumer involvement, we ask to

increase the consumer representation on the committee and as we look in the future. And the public comment is great. But time for dialogue is always much needed. And we'd like to see the ideas for the -- to collaborate with groups for information and data collecting. When we talk about needing that evidence research and the numbers, it's really to tap into those groups and get the numbers. The numbers are there that we are looking for, in some ways. And they're great to help guide to find greater numbers.

To get more consumer involvement here -- I know budgets are tight. But (inaudible) need something to possibly look for more scholarship funding to get folks in from across the country who do not have the economic means to be here but would really like to be a great voice for their disorders and their needs.

To continue partnering with consumers and advocacy groups with committee initiatives like the clearinghouse and representation with the regional collaboratives -- this is huge, because it is a great way to utilize the consumers who want to be a voice,

but cannot make it to Washington, D.C.; and encourage providers to link newly-diagnosed patients and families to advocacy groups to begin that collaboration from day one.

Unfortunately, we have parents of children who were diagnosed at birth, but are just now finding their organizations to tap into support and information sharing. And they live a life of isolation. And in 2011, we don't need to have that. But it's a partnership, and it's communication sharing that has to happen with the medical profession as well.

The advocacy groups and the nomination process to help move that along -- we know that will continue. They are great resources submitting their nominations for their disorders that are to be considered. And they come with providing very disorder-specific information from a different perspective that might not be in all the evidence review.

They are a great entity to have participate in the evidence review work group discussions early on

that, maybe, will help in looking and addressing the evidence review issues. And the consumers of disorders yet to be included on the recommended panel are really critical stakeholders. They are the people that are still losing their children. They are the consumers that are still looking for that service to help make that change.

These stakeholders, they understand the difficulties and the numbers. And the reality that the great numbers that, as the discussion earlier heard, they won't exist. We won't have those great numbers. But every life that is diagnosed with one of these conditions is very valuable. And they are a statistic. And we'd like to see, over time, to have less statistics of these children still dying from their disorders, but rather being able to join the panel making a difference.

In looking at the consumer viewpoint, one final comment is that the adoption and success of newborn screening and related issues is really going to depend on whether the needs and concerns of these consumers and advocacy groups are addressed and

harnessed as a driver in the medical profession and public, or whether they will lead to some apprehension and distrust from the public stakeholders. I think we've already started to experience the low effects of some negativity of mistrust from some entities about newborn screening, which is something that we all want to really protect and preserve what we've accomplished so far.

But we all recognize that there is a lot of work to be done. And it's not going to be so easy with these new disorders that are coming down the pipeline. And consumers really understand that, but really want to work with the committee to really help overcome the barriers there to find a good, cohesive way to overcome and make those challenges -- to rid them and really, possibly, find a way to meet everyone's needs and help those consumers find that entity that really are looking for.

And what it comes down to, at the end of the day, when looking at all of this, the successes of this committee, I had to put up here, translates into a child's future. And this is a little girl who was

one of the newer diagnosed children after newborn screening was expanded. And the committee recommended the panel.

And this is a little three-year-old now who's going to preschool this fall. She just started last week. So the work of this committee has really enabled this child to now have her future the way we all hope for children to have.

And we hope that the work will continue so that we can continue to see more cases like this and have -- you know, living out their lives. So I just thank you. And that is my work. And I just am -- I applaud this committee from day one and am really proud to have been a part of it. And I wish you the best in continuing to address these really difficult and complex issues. Thank you.

DR. HOWELL: Jana, thank you very much.

(Applause.)

DR. HOWELL: I don't think we can underestimate the extraordinary value of the advocacy community in taking recommendations from this committee and making them happen at the local level.

And I think that it's been very gratifying to see the advocacy community, such as Jana, take the recommendation of this committee into their plans when they're advocating so that they're advocating for conditions and programs that have been thoroughly vetted, and so forth. And so, we are very grateful.

Any questions or comments for --

Alan?

DR. FLEISCHMAN: Well, I do want to echo, Rod, your comment, because I think the advocacy community of patients and families are critical, particularly in the present environment of fiscal constraints on departments of health out there in every state. And I think that we may want to consider, as one of the future activities of the committee, to understand those implementation constraints and difficulties at the state level, because, as this committee makes its wise decisions and the Secretary adopts them and helps us dramatically with her recommendations, we find that, at the state level, every one of those states is in dire straits and is working very hard to maintain,

never mind expand, the kinds of work that they do.

So I think this committee may want to do that. And I think our advocacy supports will be absolutely critical in those state-by-state fights.

DR. HOWELL: I agree.

Tracy?

DR. TROTTER: First, in full disclosure, I will have to say, because I'm sitting next to Ned, I have to say that I've had the pleasure of having Jana as our Co-Chair for our subcommittee for the last four years, and having Andrea Williams, who's going to be joining the committee in January, as a member of that subcommittee. So I've had more positive opportunity to find out how well this system works than usual.

The second is that I'm in general pediatrics, so I actually spend my day seeing children and their families with special health care needs. And so, I think it's important this 20 minutes refocuses what we do. The end user, if you're selling something, using Jana's consumer report, the end user is the patient and their family. The client, if you're a lawyer, is the patient or family.

For physicians, it's a patient. And patients and their families are what we do. And it's what we're here about. And it's the end result of everything we do, is that picture. And I really appreciate Jana bringing that into focus. Thank you.

DR. HOWELL: Let me comment, make one other comment, about the folks in the audience at this meeting. It's been very gratifying with the very large attendance that this committee has routinely had. If you go to most other federal agencies and committees like this, 10 seats would be added, but with some vacancies. And so, to have this large group of people who have been active and interested and helping make things move along, certainly, the committee has been very aware of that. And I have personally appreciate that a great deal.

Any further comments?

While we're wrapping up this session on some past history, and so forth, it's important that I acknowledge the extraordinary activity and support of Michele Puryear, who, as you know, was the original Executive Secretary of this committee and served in

that role until the -- through the 24th meeting. And much of the activity of this committee and the organization and making it move along wouldn't have happened without Michele.

And I think it's very important that we recognize her contributions and wish her well as she's currently in the Office of Rare Diseases at the NIH. And we hope that that office will soon be expert in newborn screening. I'm sure they are. They're hearing about it day in and day out.

Are there any other comments, and so forth?

Let's take a break. And we will return at a quarter of 11.

(Break.)

DR. HOWELL: Ladies and gentlemen, I think we should start.

Chris Kus needs to sit down.

Mike Watson needs to sit down.

Jane Getchell needs to sit down.

And who else?

And then, everybody needs to stop talking. We're going to now move into a section that we entitle

the present work of the committee. As you know, the committee has -- we've been talking about a lot of the activities of the committee, prior to our break. But we, obviously, don't function in a vacuum, and we have many important partners that support the committee.

The committee's charged the Education and Training Subcommittee to start a newborn screening awareness campaign. And in order to conduct the campaign, a scan of the current status was determined by the subcommittee to be the first step.

And this committee, through our contractor, which is Altarum, who does the committee meetings, and so forth, subcontracted to have a media scan completed. And we're going to hear a report from that. And it's going to be a newborn screening awareness campaign report on the media scan. And our presenter will be Jennifer Nichols from the Porter Novelli Group.

Thank you very much for your wisdom. We'll look forward to hearing you.

MS. NICHOLS: Good morning. Thanks for having me. So I'm Jennifer Nichols, and I'm here from

Porter Novelli and happy to be with you this morning to share a little bit about what we learned during the environmental scanning process for newborn screening. I'm going to leave you a mystery for a moment.

We are working with Altarum and HRSA on a phase one to a potential newborn awareness campaign raising awareness about newborn screening. We have a three-step process to that. And our first step is environmental scanning, which is a broad process of learning what's on the Internet, what are health care providers saying, and what is actually reaching consumers.

We then go to a deeper dive in the people who know what's really happening in the newborn screening field and do a strategy from it or some form of partner consensus-building meeting to incorporate both what we found out that consumers are seeing and what's happening actively in the field. And from those two pieces of information, we will come up with recommendations for how to proceed with a newborn screening awareness campaign, what the next steps might be for that.

I think he's coming back. So I'm just going to keep going. And then, you'll get some surprises with my slides, I guess.

So we start the environmental scanning process in a very broad way. We use a guided approach, but we call it guided with a little bit of exploration. So we start with standard search terms. It's primarily Web-based.

We're good? Ta-da. Okay. Let's catch up. All right. Here we are.

And we use, kind of, a "see where it leads" approach. When we approach our environmental scanning, we're looking at it more from a -- if we were a parent to be, a parent, or perhaps a mother of a new -- someone who's about to be a parent, and I wanted to find out about newborn screening. Where would I go, what kind of information would I look for?

So it's important, as you're hearing the results that we found, to keep in mind that this is the lens. We are not doing a traditional literature review. We're looking at it from a "if I went on Google, what would I find"? And then, we take it a

little bit farther. Most people don't go to the fourth page of Google search results. But we go that deep. We look at Yahoo and Wikipedia and WebMD. So we're really searching across a Web medium.

And there's a reason that we do that. We have a proprietary database at Porter Novelli called the Style Survey. It's licensed by CDC and other agencies within HHS. It's an annual survey to get consumer perspectives on different health issues. And this is from the Health Style survey from 2010.

And, as you see, the doctor and the Internet are the most popular places that people go to when they're turning for help information. So during this phase, we were not actually speaking directly to health care providers, but we did look into what are health care providers giving to their patients as well as most of our time was spent on what are people finding on Dr. Google.

So I'm going to talk a little bit about each piece that we listened to. And, again, first step are what are people Googling. We know that this is what consumers go to now, is they want that first hit of I

have never heard of newborn screening, what does that mean. Go Google it.

Google is actually a verb. So we looked at Google, Yahoo, as I mentioned, WebMD, Wikipedia. And we used a standard set of search terms across all of these to pull up what might people find if they look for newborn screening or heel prick test or other words that they might have used to try and figure out what this is.

And, as you can see, the most frequently referenced sites are CDC, the American Academy of Pediatrics, and the March of Dimes. Other sites that are coming up frequently, but not as frequently include NIH, the Cystic Fibrosis Foundation, Wikipedia, and WebMD.

So when we looked a little bit deeper and found, okay, this isn't just popping up frequently, but what is it actually putting out there. And what we found consistently across the most frequently referenced sites was that it's very education-focused. So it's giving the basic definitions.

It's talking about health impact, both for

an individual child and for society overall. It's looking at the benefits of early diagnosis and treatment, talking about how it varies by state, and talking a little bit about how the procedure works and the timing. So it's very information-focused. There does not appear to be a bias positively or negatively on these sites. It's neutral information-driven.

We also went and observed specifically, as we could from a secondary approach, what are hospitals and health care providers putting out there about newborn screening. Because we weren't talking directly to them during this phase, we used their Web sites. And hospitals are actually providing more significant information on newborn screening than an average pediatrician Web site.

Pediatricians often have links to the American Academy of Pediatrics and the American College of Medical Genetics. But hospitals have those links as well as some specific information about different conditions that are being tested for, or screened for, excuse me, and the explanation in how it varies by state. So this is looking at the specific

hospital Web sites across different locations.

Once we did, kind of, a broad sweep of what's on the Internet that consumers might be seeing, during the month of August -- so we concluded this process about August 30th -- we looked at the media audit. And this spanned back about five years. We found about 300 unique articles that got pulled up from different media sources, whether it was newspaper or broadcasts or radio, about newborn screening.

When we actually, kind of, sifted through those and saw what is the main topic here, there were only 88 that were actually really relevant to newborn screening. So some of the tests -- some of the search terms we used were things like genetic tests or heel prick test or just screening in general. And those would pull up other things that weren't really actually related to newborn screening.

So only about 30 percent of the articles that we found in our search were relevant to newborn screening. That's 88 articles over about a five-year period.

They ranged -- whether they were coming from

the Web or newspapers, there wasn't a consistent source that was really publishing more information. They were both national and locally-focused and looking very similar to what we found on key Web sites. They were education-focused. They were mostly neutral or positive in their messaging. And there was very limited press on the negative aspects, or perceived negative aspects, of newborn screening. And there were many articles on disease-specific issues.

Using Google alerts, which probably many of you have -- it's a great tool to keep on what's happening out there in the media world -- we got a heads up that it was Newborn Screening Awareness Month. So even though we concluded the actual media audit search, we went back and looked.

And Newborn Screening Awareness Month was getting hit in the media over the first two weeks of September. And, again, the information is very simple, basic education information and primarily has a positive spin to it. So it's focusing on the benefits of newborn screening.

Beyond what is very intuitive, first nature

for consumers to look at, whether it's on their TV or on their Internet, we also know that specific organizations influence what parents are looking for, especially the American Academy of Pediatrics for new parents or soon to be parents. We found a great list of stakeholders that have specific information for consumers.

And I want to point out here that this was a very targeted search to look for organizations that provide resources. This was not the same method that we used through the consumer lens. So this is our actually trying to find out what's out there that's available, but not necessarily what's popping up on the first four pages of returned search results on a Google page.

We also looked for campaigns specifically that had been done to see what was out there in the field that were, kind of, broad, sweeping messages around newborn screening. And we found one comprehensive campaign that had been conducted by Saving Babies Through Screening and two campaigns that were very specifically focused on one condition.

So, again, this is a we're trying to learn more. This is not what a consumer would necessarily find if they typed into Google. But we did want to find out what's out there in the literature that can help us understand what that consumer perspective is and what they might be thinking about newborn screening.

So we did go to the literature and look specifically for that attitudes and perspectives that parents may have related to newborn screening. And we found that it's generally positive. It's just perceived as part of what happens in hospitals. There's a little bit of anxiety about what happens with a false/negative or a false/positive result.

A lot of the conditions are not something that are familiar to consumers. The names are not things that are common to them. But there are a few things that are more familiar that are being screened for, like sickle cell. And that, overall, there's limited knowledge and understanding of the issues of residual storage and research related to the newborn screening process.

So, in summary, I whipped through this, because I thought there might be some questions. We found that overall, there is information online about newborn screening, but that it's only moderately accessible. The information that is valuable to consumers has not been optimized. It's not necessarily readily available to an average parent to be going out to search for new information.

The messages at this point appear to be primarily neutral or trending towards the positive aspects of newborn screening. Media and campaigns, which would be how we would talk about consumers being indirectly exposed or not necessarily looking for that information specifically, very limited in what's happening in that indirect exposure.

We don't feel confident in really talking about what health care providers are providing to their patients at this point. We know it's on their Web sites, but that's such a very limited piece of how patients interact with their health care providers that we really feel like we need more information on that front before we can speculate much about it.

So, with this information in-hand, we feel like there are definitely some big pieces missing here. So having the consumer perspective in doing an awareness campaign is absolutely critical to how we would approach raising general consumer awareness. We need to understand where they're starting from.

But there's also a really important piece of knowing what's going on in the field and how what's already happening can fold into an awareness campaign. So next up on the phase one approach that we have is doing a consensus-building meeting with partners and other stakeholders to come to some good recommendations and next steps for proceeding with a newborn screening awareness campaign. And that's it.

DR. HOWELL: Thank you very much.

Are there questions of Jennifer?

Jeff?

DR. BOTKIN: This is a wonderful project, very interesting. There is some literature out there. And Terry Davis' group, for example, did a number of focus groups five or six years ago, sort of, to find what parents want to know about newborn screening.

And I think one of the key outcomes of her research was that parents don't want to know nearly as much as we're afraid they want to know about. In other words, they don't want to know what the list of conditions is and that level of detail.

So I'm wondering whether part of the project is, sort of, assess these sites by those sorts of criteria. Do they meet what we think we know about parents' educational needs about this topic? Or are those elements, sort of, embedded in a much more complicated data field that might be challenging for people to navigate?

MS. NICHOLS: I think that's a really good point. And one of the things that we found in doing just a very standard Google search with all of our search terms was that there are a lot of things that are popping up that are not relevant. So we screened this with an eye for what is it that we're looking for, knowing we're looking for newborn screening information.

But in that field that's popping up on those first two to four pages of Google search results,

there's all kinds of stuff that is not relevant to a consumer mixed with things that might be very relevant in the content, but in the delivery is not something that they're going to necessarily digest or want to read through. So even though if you're in a college course or even a high school course these days, Wikipedia is not an accepted reference for paper writing.

Wikipedia and WebMD really are sources that people go to, because it's easily digestible information. I think that's a good point in balancing that, what's available versus what consumers really comprehend and take in.

DR. HOWELL: Alan?

DR. FLEISCHMAN: And thank you for this really very important beginning of this project. One of the things I'm struck with, though, in that last bullet -- most families don't come in contact in any meaningful way with pediatricians and hospitals before delivery, or at least before labor or before induction, even though they shouldn't be having all those inductions.

(Laughter.)

DR. FLEISCHMAN: But they -- I needed to say that. I needed to say that. My people are here.

(Laughter.)

DR. FLEISCHMAN: But the point being that the obstetric community has not embraced this educational activity, neither the nurses nor the obstetricians, for good and important reasons. And they're not at the table today, which I'm always upset about when they're not, because I think they're an important part of our educational armamentarium. And so are the nurses.

So I hope that, as we think about this, while the American Academy of Pediatrics has done its job, it's not getting to the parents at the time when they need the information. So I think we need to keep that in mind in terms of educational activities.

And although ACOG has, on their Web site, a whole bunch of stuff about newborn screening and tells their obstetricians they're supposed to educate women about that, I would doubt that we could empirically measure a universal exposure to such education.

MS. NICHOLS: Thank you. I think that's a really important thing to note. We'll have to update that last bullet. And I will say that what we found on ACOG's Web site was more focused on the materials for physicians to talk to patients as opposed to materials to pass through to patients.

DR. HOWELL: Sharon?

MS. TERRY: Great report. How will you address the fact that what we're, sort of, looking at is a snapshot of the past by looking at Web sites and professional societies, et cetera, because parents to be are going to seek and consume information in very different ways than -- and they already are, actually. And research shows that -- than we have traditionally -- and to say that Web searches are traditional sounds really crazy, but they are.

MS. NICHOLS: Right.

MS. TERRY: So how will you address being ahead of the curve, if we do decide to go out in a campaign, kind of, mode?

MS. NICHOLS: I think that's a really important question. Something that we have wrestled

with is when 95 percent of the media that we're seeing is neutral or positive, it's really hard to necessarily justify, well, of course, you need to raise awareness. If everyone feels pretty good about it, what's the point of that?

But I think, in addition to people changing the way that they get information, there's also the potential really quick turnaround in information that's available. And the media cycle can, you know, immediately turn that on its head. So it's figuring out that balance of where does an awareness campaign fit.

And is it something that you can get out there ahead of time to reach consumers with? Even if nothing bad ever does happen, you still want it out there. So I think that that's a good question. You know, being a researcher, I always say, we need to talk to parents more. And I think that that is a piece of it, of learning how they get their information, but also really figuring out how do we put the right information there. So --

DR. HOWELL: Alexis?

DR. THOMPSON: I think it's a phenomenal project. I had a question regarding patients or families or communities where English is not their first language and also communities where there's a high rate of poverty. When I think about, for instance, in Hispanic communities, they often utilize the radio for many of -- much of their information. And then, sort of, wondering when you got your information about what families prefer, what was the ethnic or socioeconomic breakdown of that?

Similarly, I still am often struck by how few of my African-American families have a computer at home. And so, yes, they may go to the library. But, clearly, it will require an extra effort for those families to access things that are on the Web. And so, I'm wondering were there representation in those groups in your research? And do you have thoughts about how to reach those communities as well?

MS. NICHOLS: So it's really important to note that, obviously, this assumes that it's the population who has access to the Internet, because there are many people who don't and even those who do

who don't trust it and don't turn to it. It's a safer representation of the general public now than perhaps it even was two to three years ago.

We also know that a lot of information that's on the Internet now is being sent to mobile phones. So people who don't have a computer, but do pay for their phone service might be accessing the Internet that way. We know in the Hispanic population, there has been a large increase of using mobile as opposed to a standard computer to access the Internet.

I think one of the difficult pieces of looking at an environmental scan is it is very secondary. It is hands-off. So we're looking at what's available to us. And we're not talking directly to people yet.

With communities that don't use the Internet and perhaps aren't accessing their health care providers, there's a huge word-of-mouth component. And finding out what that word-of-mouth is takes a totally different approach, which we would indicate would be, kind of, a phase two. Once we figure out

what we're trying to get at, who do we talk to, and how do we get that information from them?

DR. HOWELL: And, Tracy?

DR. TROTTER: Yeah, a little background for the committee and the folks in the room today who may not know. This has risen from a long-standing concern for this need.

In fact, when I scanned the minutes of this meeting, the first time it came up was 2004 from Dr. Rodney Howell, who said, "A good idea would be a national newborn screening awareness campaign." And it became -- and I give Coleen Boyle and Angie Colson from the CDC kudos for picking this banner up about a year ago and then came through the Education and Training Subcommittee, as you know.

And what was conceived was a four-phase program that would, in many ways, attempt to replicate in some way the autism campaigns, the back to school -- back to sleep campaigns, the immunization campaigns that have been very successful in the United States and in maintaining a positive view on public health matters of importance. And what was approved by all

of us here as a committee as a whole was the phase one. And this is the beginning of phase one, is to get this scan.

The end of -- the second part of phase one is to try to bring as many stakeholders together as is possible to get a little more real-life look. And Alexis brought up some good points there, that we need to know those other pieces to then, hopefully, bring back to the committee as a whole, is this feasible, should we move ahead with phase two, how should we do that. So this is, sort of, the opening salvo of approaching this as a potential campaign in the future.

DR. HOWELL: Fred?

DR. CHEN: I just wonder if part of your analysis is going to -- you know, one of the realities of newborn screening is it's different from many of those awareness campaigns in that it's already very successful and near universal. And I wonder if one of your analyses might be potential downsides. You know, even though most of the coverage is predominantly positive, what do we stand to gain when there's

already a near universal uptake? Is there a downside to raising awareness in that potentially parents will start to opt out?

MS. NICHOLS: So I think that's a really valuable question. And if we knew that the media and Internet landscape were going to stay the same, the answer might be that the value is not worth the cost. I think one of the things that we learned -- Porter Novelli worked on the Learn the Signs, Act Early campaign, which is about raising awareness for developmental milestones. But it started as an autism awareness campaign.

And one of the things that we learned in that process was that we didn't get out ahead of the message. And the message was already forming itself. And we needed to address what was out there instead of just focusing on the issue itself. So that seems also our -- I think you could argue a public health success.

But they were threatened with many messages coming from the media and, at that point in time, coming from the literature. So I think part of asking

that question is also saying -- I mean, as Sharon noted, this is a point in time. And at this point in time, it looks like we've got positive and neutral messages reaching parents. But as we look to the future, figuring out what are we trying -- what can we potentially project might change, and how do we address that.

DR. HOWELL: Katherine, do you have a quick comment?

MS. HARRIS: Very quick comment. NYMAC is working with Genetic Alliance spearheading and talking about providing information to parents to be working with childbirth educators: doulas, midwives, those people who are teaching women what to do when they have a child and giving them information about newborn screening. So we're working on getting that program started.

MS. NICHOLS: That's great.

DR. HOWELL: Thank you very much.

Don? Okay.

DR. BAILEY: So, you know, I think I'm very much in favor of public transparency and public

awareness. And we need to do it in a very intentional kind of way. I think the kinds of things that Tracy was just talking about -- most of them had fairly clear objectives about change that you wanted to have happen.

MS. NICHOLS: Right.

DR. BAILEY: You wanted to get babies sleeping the right way. You wanted to get kids screens more like they'd be screened for autism. So I think that would be key to this campaign in the next phase, is not only figuring out what the messages are, but what are our goals, what do we want the messages to accomplish.

MS. NICHOLS: Thank you. I think that's a big piece that we look to hope to achieve from the strategy and the consensus-building meeting. Thank you.

DR. HOWELL: Thank you very much, and so forth.

We're going to move ahead now. When the Advisory Committee was reauthorized by the Newborn Screening Saves Lives Act that we've heard about

already today, in this legislation, there were many projects that were outlined. And we'll hear about some of these partners next.

And the first person on my agenda is Sharon Terry, who will give us a tour of Baby's First Test. And Natasha Bonhomme is also listed on the program. And here she comes to assist in some very effective way, I'm sure.

MS. BONHOMME: Actually, (off-mike). And (off-mike). So we wanted to actually start with some questions (off-mike). We wanted to start with some questions, which is an odd thing to do, perhaps. But we thought we should put these right up front, because our way of engaging in this project, as Rod says, which is required by the legislation, is to really engage the community in multiple ways.

And, as you know, there are multiple audiences and multiple communities. So some of the questions that you will see -- you will see these questions. I mean, they're not written on the screen during the tour, but they will pop into your mind -- are issues around the recommended universal screening

panel and other conditions and how should they be included in the educational efforts, what's the proper language to represent the all -- all of the states required detected mandated.

As we all know, the states have different language, different ways of expressing things. And we want to be sensitive to that. And we want to be able to provide a cohesive message to the public, very much building off the last presentation.

Terminology -- what terminology should be used as the reference point. And there's a number of terminology recommendations out there. They are not harmonized. This is not an unusual or specific to newborn screening issue. It's one that's pervasive across all rare diseases. And anyone can look at (inaudible), Office of Rare Disease Research, Orphan Net, Mesh and just see the, kind of, myriad of ways that people express the same condition in multiple ways. So it's another area that there is broad discussion around and that we're going to pay attention to.

And then, key messages -- are we looking for

awareness simply through the site? Or are we looking for informed decision making through the resource? And those are, kind of, two different ways of looking at something like this. And again, pertinent to my question to the last speaker, it's a critical time for us to understand that the communication tools that we've been using are evolving.

And so, something as simple as a Web site when it was Web 1.0 as it becomes Web 2.0 and becomes engaging and it becomes Web 3.0 and actually becomes empowering and part of my decision making matrix as a person, how are we going to reflect that in Baby's First Test? So I now turn it over to Natasha, who will drive you through Baby's First Test.

MS. BONHOMME: Great. Thank you. I get to do the fun part. So this is Baby's First Test, which is up and running. And this is meant to be the nation's newborn screening clearinghouse of information. I'm going to just go through some of the highlights of the site. There's a lot that I could (inaudible) in detail. But I'm really just going to highlight some of the key things and then have time

for questions.

As you can see here, this really is how the information is laid out, with the general information about newborn screening, where it goes into just general screening facts, resources, also genetics and family history. Again, the key point of this site is that people could go and get as little or as much information as they want.

As was mentioned earlier today in the presentation just before, some people just want to know the very basic information. And then, there's some people who will really want to be able to drill down and get a lot more nitty gritty. So we want to be able to provide that in an easy-to-navigate way.

The next section here, which is what to expect -- we start with before birth. And I will click on that just so that everyone can get a sense of what type of information we have there. But we want this conversation to start, really, even before women are in the hospital getting ready to deliver. So we talk about the seven things parents want to know about newborn screening, which is based off of the HRSA-

funded project of the same name, Seven Things Parents Want to Know.

We also go into more detail about screening procedures. We also talk about results as well as different screening outcomes and what happens to the blood sample. The reason why we laid it out this way is that these were the key questions that we felt people would come up to and would want to know about.

I'll click on screening procedures just to give you also a sense of the site and how it's laid out. So generally, each section has an "in this section," that really talks about some of the key points that are on that page. You'll notice, going through this, that the pages are very long. There's a lot of information. We'll be doing usability ability testing to see how people would like that information laid out.

The reason why we laid it out in long pages is, actually, because we found that when people don't even know what they're looking for, if you just collapse it into headings, oftentimes, they'll just skip over it, because they don't realize that's a key

piece of information. And especially since we're talking about newborn screening, something that many people don't know about, we wanted to make sure that people did not skip over information. So, like I said, we will be doing some more usability testing about how that's laid out.

Also, (inaudible) this year with either key questions, other resources. Again, wanting to be able to give people a number of different opportunities to educate themselves, but not necessarily bombarding them with just a laundry list of links.

If we go to living with conditions, we also want people to be able to use this site once they actually do have a diagnosis. We thought it was really important to be able to highlight the family experiences and also some of the other issues that may come up after a diagnosis.

So if we go here, we talk about family experiences, how do people talk about a diagnosis, advocacy and support groups, finding a specialist, insurance and planning, and looking to the future. And these items were really brought to light based off

of the research that we did through our consumer focus newborn screening project, which we worked with the Genetics in Public Policy Center out of Hopkins as well as the University of Maryland. In terms of these are just some issues that come up, both during that diagnostic period and then, when someone actually has a diagnosis.

So we can click on the family experiences. One thing we wanted as a key message throughout this site is it's important to get follow-up, it's important to really speak with your health care provider, and if you actually do have a diagnosis, that there is, kind of, life after that and that, because of newborn screening and because of the interventions, people can have really, kind of, fulfilled and really have healthy lives. So that is what most of these videos currently showcase.

So let's go back to the home page. So the layout -- again, this is, kind of, a faster way to get through some key information. We'll go into state programs in a moment. People can also look up their specific condition here.

And then, we also -- from the beginning of this project, we've been very interested in terms of social media and the different ways of engaging a conversation around newborn screening. And this bottom section here really highlights that.

We have our "in the news," which we keep fairly current in terms of some of the major things happening. We have our "blog," which is updated once a week. We are looking to do the front type of blog partnerships. We, actually, in October will be doing one with the American College of Nurse Midwives, where we will do some cross-posting, again, to get the word out to another group of people who have contacts to parents.

Our "community corner," -- of course, this week, we would be highlighting the Advisory Committee. But let's say you want to see what's going on in a state program. I'm sure many people are interested in that. So you would click here.

And we're looking to see how we can make that map on the front page actually clickable, based on the different states. But right now, either you

click there -- and you automatically go to a section that talks about what is a panel. And we go into some detail about that, because that is one thing that, if you are not in this community, you may not know what is a panel. And we go into some information about that.

But let's say we go to New Jersey. We want to see what's going on in New Jersey. And one thing Sharon had mentioned in terms of some of the questions that we are looking at is really what is the best way to represent the information, particularly the conditions that are screened for. This is always a conversation that many groups have had different issues around in terms of speaking or writing, listing out the conditions.

So what we've started off with -- and this really is just a foundation. We really do see this as an evolving project that there will be different iterations of. But we really started with the RUSP, the Recommended Uniform Screening Panel. We felt that that was a good starting off point, particularly based off of just information or feedback we had gotten from

the Secretary's Advisory Committee as well as trying to find what is a good way of actually showing the uniformity across states.

One of the questions for the past two years people asked is how are you going to make sure people aren't going to the sites just to compare states. And that has never been the intention of the site. It really has been to how do we highlight all the good work that's being done, the newborn screening state programs, and getting the word out.

So again, here we have the contacts. These were all pages that were sent to the state programs. And we did get their feedback. We're still getting feedback on that. Again, this is really a living Web site in terms of its evolving every single day.

Then if a state had specific resources for health professionals, we would put that here. If they had a specific brochure for parents, we would also put another box linking here. Again, long list. And this is something that we will be looking to get more feedback on in terms of how do people really want to see this information, again, building off of the work

that Terry Davis did.

And then, we just go into some general information. The program overview -- we did want to include how newborn screening is paid for, particularly because in this type of economic crisis, if you will, newborn screening isn't free. And we wanted to be able to highlight that. Even if families are not the ones directly paying for it, we thought that that was a key message in terms of being able to preserve the budgets of the state programs. And the only way to highlight that is to say it actually does cost something.

We have some opt out resources, the support for families. In this, we will be expanding this to also include the family voices chapters of the different states. But this is really just, kind of, a preliminary go.

And then, also storage and use of DBS, which we will be changing to residual dried blood spots since not everyone knows what DBS is. But that is the general layout of what all the state pages look like.

So the last place that I want to take you

before taking some questions is "find a condition." So let's say you want to see sickle cell. So I would type that in, and you would see that it starts to automatically populate.

This was something that we thought was really important, because different people may -- someone may have said it's a hemoglobinopathy. Someone may have heard that it's sickle cell. We wanted to be able to make sure that we cross-referenced that. So all of these conditions are cross-referenced in the back end of the site.

So now, here we are at our condition-specific pages. We do have a section that says "also known as," again, addressing the issue that different conditions are called different things by different state programs. This is another area where we're really eager for some feedback. What's the best way to represent this information without confusing people?

We have our "at a glance," so, for people who just want a very quick snapshot. We also have information that's specific to health professionals,

highlighting the act sheet.

And then, we have just some general information about the condition. All of these pages were actually sent to the main advocacy organization of that condition to be able to get their input also. We wanted to make sure that we were being very representation and in alignment with the key advocacy organizations.

If we go here, there's "early signs, treatments, expected outcomes." And again, there's a lot of information here, but it isn't as if you have to read through all of it. It really is in a tiered fashion.

And we also have our "support services, access to care." So "where did we get this information"? And this is a link to all of the resources that we lent to to get information. We have the Star G program. We have National Library of Medicine, ACMG. So this really does show, kind of, where our evidence came from.

As I said, I could probably go and talk about the site for another hour, but I know we have a

number of other presentations. So I'm happy to take questions at this point.

DR. HOWELL: Thank you very much, Natasha.

Joe?

DR. BOCCHINI: I think you've created a remarkable resource. I think it's a really wonderful job.

I guess, two questions -- one, reading level -- if you, sort of, target a specific reading level for the parents. And then, two, other languages -- are you working on that as well?

MS. BONHOMME: Great. For reading level, generally, what we call the primary and secondary navigation, so, really, the general newborn screening information, so what's highlighted here, that we have aimed for it to be at about an eighth grade reading level. We will be going back and doing a literacy review to try to bring that down even further to potentially assist the sixth grade reading level, since we know that that is actually the average in this country.

For the condition-specific pages, that's a

little tougher, because then, you know, we're getting into the big words. But that is something we're looking at.

In terms of other languages, that is something that I would like to see, at least at the end of this project year. And our project year goes from September to August. So by August 2012, we are really looking to see if we can have the site in Spanish.

The main thing is is that we didn't want to just put a general Google translator, because there is so much information here, and a lot of it does have to do with medical or health issues that we didn't want to inadvertently, all of a sudden -- confusing a different group of people in a different language. But that is something that we are looking at and looking to see would it be best to just focus on the general newborn screening information, translating that first and then, in a second phase, translating the state-specific and condition-specific pages or doing that in a once-all swoop.

MS. TERRY: I'll add a little to that, too.

And the funding for the site from HRSA doesn't include much money for translation. It was very expensive to do interpretation and translation. And so, one of the things we'll be also doing is looking for other funders who are interested in specific communities that would need this information and be interested in funding those kinds of interpretations and translations.

DR. HOWELL: Thank you, Sharon and Natasha.

Quick comment?

MS. GYREN: So when you Google -- I know it's old-fashioned, Sharon, but newborn screening and opt out, I'm speaking about that section. You get, you know, Minnesota. Okay. So I'm just wondering how you -- sort of, where you are on that, since you do have a section on opt out.

MS. TERRY: So, Nancy, do you mean where we are on having this information rise to the top of Google?

MS. GYREN: Yeah.

MS. TERRY: So we, actually, have, in addition to the literacy stuff and some other reviews

that are going to go on this year, also optimizing for search engines. And that changes quite frequently. In the old days, it was as simple as making sure our embedded meta tags said opt out, for example.

And now, there is a whole bunch of other characteristics. There's actually 12 of them that we're carefully monitoring throughout the site to make sure that it has the right links in and links out, that sort of thing, to rise to the top.

The tough part with that, of course, is everyone is working on those analytics and metrics. And so, other sites are doing the same thing. And one can never guarantee where one would come in Google.

The other part of that, though, is Google and we have a relationship, since I served on Google's health board. And Google -- we are going through the vetting process of being one of their trusted sources.

I don't know if you've noticed, when you Google a disease, there's a bunch of information that comes up at the top that looks like it's separated. And they have things like Mayo and Kaiser that they've vetted and decided those are good sources of

information. And they're looking at us for that right now.

MS. GYREN: (Off-mike) opt out?

MS. TERRY: Yes.

MS. GYREN: What's your message?

MS. TERRY: The message for opt out.

MS. BONHOMME: Say that again, just a general message in terms of opt out from the site? So really, what we're seeing for that is that that is something that you really should discuss with your state and with your health professional, that there is a reason why there is newborn screening. And that's one reason why all those opt out sections did go to the states themselves, since every state does say something a little bit differently.

Some states said they only wanted it to be in relation to a religious opt out. And then, others said just wanting to give more information. It actually goes back to that third question that Sharon posed in terms of the difference between awareness and an informed decision making. And that is something that we'll continue to work on.

And I did just realize that the site was actually much bigger on my screen than it was on here. So you guys in the back probably didn't see it. So I apologize for that. But it's baby'sfirsttest.org. And you can definitely send questions directly to me about that.

DR. HOWELL: Thank you, Natasha and Sharon. And we will see you tonight.

MS. TERRY: Yep.

DR. HOWELL: At your festivity.

We're going to now move to the Newborn Screening Translational Research Network. And we'll hear from Mike Watson, who is the -- obviously, he's ACMG representative to this committee and the Executive Director of the American College of Medical Genetics, that holds the NICHD contract for the Translational Research Network --

DR. WATSON: It does.

DR. HOWELL: -- Coordinating Center. I sense the need for speed coming here.

Yes, we do have the contract from NICHD to develop the Newborn Screening Translational Research

Network. Let me -- which one of these is going to move the slides? All right.

So you've seen this slide already. Alex showed it when he presented earlier in the context of what it includes around the Advisory Committee's activities. But in the same Newborn Screening Saves Lives Act is legislation that established the Hunter Kelly research program at NICHD.

That is broadly the Newborn Screening Translational Research Network activities of NICHD for which we at ACMG operate the Coordinating Center. And we're now in a phase where we're moving from what we've been doing centrally to integrating grantees and contractors into the infrastructure and resources that we've been developing. And that's what I'm going to try to walk you through pretty quickly.

Really, the goals are stated in that Newborn Screening Saves Lives Act. They are to capture the evidence around newborn screening activities, particularly the conditions that are candidates for newborn screening, conditions that are already there that may not be as well-understood as we would like,

because we began to really understand them when we arrived in newborn screening with these conditions.

So the kinds of research that is envisioned to operate through the Translational Research Network includes assessing new technologies that might be applied to newborn screening, assessing new conditions that are candidates for newborn screening. This includes supporting the pilot studies that take place.

We know there's enormous variability in the number of babies born in different states. And with these rare diseases, it was very clear that, to understand them well, we needed to figure out how to play together across multiple states to really pull the data together in a much more rapid way to get robust information as quickly as possible. And that can only be done through relatively broad collaborations.

And we've already alluded to severe combined immunodeficiency as an example of how much more rapidly we were able to capture data and move along. And I'll touch on that only briefly in a little bit.

The first wave of grants that were awarded

by NICHD in the program were in the area of development of clinical histories of conditions, both those in newborn screening and candidate conditions for newborn screening. And I'll tell you where we are with those briefly.

Outcome studies are also important. And that's that longitudinal health care information following the diagnosis of the patient and the treatment that captures their, sort of, interval visits to the physician and how they're progressing in their treatment and long-term outcomes, which are critical to that look-back, I think, that the committee is interested in to know whether or not newborn screening made a difference or not. And we envision, as more and more therapeutics for conditions come into play, certainly, clinical trials will have a place, certainly, as they relate to that broad population impact around clinical interventions for these conditions.

So just, who we are -- I'm the Director of the project at ACMG. Barry Thompson's our Medical Director. He'll be speaking, actually, after me about

the Regional Collaborative National Coordinating Center activities. Amy Hoffman manages the project on a day-to-day basis. And then, we have people who are dealing more on the -- with the individual grantees: Amy Brower, Bruce Bowdish, who oversees all of our I.T. informatics, that crosses all of these grantees and contract groups that we work with and a number of other people who are critical to any of us getting anything done, in the end.

We started, really, in a development phase for the NBSTRN by establishing a number of committees. We have a standing committee that oversees much of what we do. That's currently Chaired by Harvey Levy and Sue Barry. We have four major work groups that, sort of, define the areas in which we anticipated we would have activity.

Clinical centers had a lot of activity to develop the data sets that define diagnosis and follow-up of patients in newborn screening. And that was something we wanted to do very early, because we wanted to integrate that with the National Library of Medicine into the standardization process for the way

you say something in health care so that it would become part of what manufacturers of EMR systems and others would be building into their systems so that ultimately, if we're lucky, we get away from this independent capture and can go into medical records to capture the kind of data we want to understand these conditions.

We have a Laboratories Work Group, which is the newborn screening laboratories and programs, who are a critical component of the Translational Research Network. And probably the most unique part of this entire activity is that it bridges the newborn screening programs in public health with the specialty providers and the primary care providers, which is a little complex and interesting, if nothing else.

We also have a Bioethics and Legal Issues Work Group that's been looking at a number of the issues that are unresolved about how we do this kind of research. And one of those, actually -- one of our grantees came in recently and hit an impediment, a significant issue, in how they might address parental permission for participating in a study where the only

way you'll ever understand the disease is to find the babies in newborn screening, because it's lethal in the first year or so of life, so to understand that we actually had to engage research early on.

Spinal muscular atrophy was that condition. And Jeff Botkin will talk more about that tomorrow, because we did a meeting on that particular topic last week.

And then, we have an I.T. and Bioinformatics Work Group that cross-cuts all of the committees, because we have to factor in the permissioning and everything else when we build the infrastructure that supports the researchers who are distributed all over the country and bring data into central data warehouses to aggregate the data from the various studies we're doing.

So the development phase included developing a Web site. We were, admittedly, slow in making that public. There was enough litigation going on that we thought it was critical that the first thing we do is generate very good information for the public on how we maintain privacy of information, how we secure the

information in our databases, and the kinds of studies that go on within the NBSTRN.

It is a resource for researchers, so we had to develop a fair bit of guidance information for new investigators and others who probably have limited understanding and knowledge of what goes on in newborn screening so they'd know what to do if they were developing their own grants to do research in this area. And the site opened in June of this year. It's at www.nbstrn.org. You're welcome to go there and look at some of the resources that are now available.

It's got both public content and investigator content. The research tools that we're developing are described there, to some extent. We've already alluded to earlier today about the need for being able to utilize the dried blood spot repositories that are out there in research.

And we've been developing a virtual repository that allows us to gaze into the resources held by those states who have been interested in participating in this program. And that -- we're really at the final stage of finalizing agreements and

expect it to open probably some time around spring to early summer of 2012.

We've also taken another resource you've seen -- the R4S Web site in region four, one of our regional collaboratives, that was used to capture data from the screening process itself in the newborn screening laboratories to help them improve their own performance of those tests. We actually have adapted that to bring pilot data in as we're developing new tests so that everybody's playing together and getting more robust data, as they progress.

And then, the tools I've already alluded to that describe diagnosis and follow-up, how we capture that at the point of care, how we move it into data warehouses or back into institutional EMR systems, and how we develop the data display tools that allow the investigators to analyze their data. And the next step will be developing the way we, sort of, bring public information about the studies that are taking place within the NBSTRN back to the public and consumers, who, without their data and information, we would not have been able to do anything in the first

place and have to be able to communicate that to the broad partnership of groups involved in making this kind of research happen.

So I mentioned the Web site. You can take a look at it at nbstrn.org. And I'm going to move along now with some quick, just, screen shots from various parts of what we've been developing.

This is the home page for the Translational Research Network. It has information for the public, for the investigators, walks people through some of the general processes and areas of concern in developing research in this area.

I've alluded to the virtual biospecimen repository. We initiated this as a virtual dried blood spot repository. But now, as investigators come in and are studying specific diseases, we're going to begin to overlay the conditions that they're studying and collecting specimens on so that we're able to extend from, not just what's in the newborn screening laboratory, but the additional specimens.

It's fully HIPPA-compliant. Secure data exchange is central to all of this. And we're now

adding in those other repositories.

And, more recently, we've decided that there's another resource that's out there that generates. It's often in industry.

They've gone to states like California and others to say, "I want to see what happens if I try newborn screening for mucopolysacaridosis, type II. So MPS II is a study that was done in California with a company. But now they have a unique cohort within their repository that we want to draw out and make visible within our own resources so investigators who may be -- or states interested in bringing those online -- begin to know where there might be resources, specimens available to move that area along.

This is the dried blood spot repository. We've been running some demonstrations and doing some functional assessments of it. You can look into the states.

You can see what positive specimens from truly diagnosed patients are available. You can see a more general population view of what's available.

And, as we begin to add additional cohorts, it should have increasing value.

There's a lot of information that explains to researchers how to use the site, what kinds of resources are there, how to search them. They can go in an see which states have, you know, the rarest of conditions. Sometimes it may take multiple states to get enough to do your research. Sometimes you might find it in a single state. So there's various ways you can parse your query of the database.

If you're interested in ruling out certain kinds of, you know, patients who might be preemies or other kinds of events that are common, there are ways of sorting through those things so that you can clean up your study population.

There are additional resources in the site that show where there are grant opportunities that relate to newborn screening, issues around state IRBs. That's a unique aspect of this, because we have, not only the academic institution that might have an IRB to deal with, but we often have a state IRB that oversees that public health function. And we are

trying to help investigators wade their way through that.

So there's a number of those types of resources that I can't show you all of them. We have means by which investigators can ask us general questions when they're beginning to think about doing research in this area. And they can then get increasingly more detailed as they interact directly with states, providing an abstract of their research and asking the state program for more information that gets much more specific about the kind of study they might be doing.

I alluded to the fact that we've taken the region four stork, or R4S Web site, that Piero Ronaldo developed for quality assurance in newborn screening. And our grantees are now using it. So one of the contractors is Dr. Dietrich Matern, who is joining this committee.

He has been curating lysosomal storage disorder component of this Web site now that's looking at comparative assessment of different technologies for screening. And we've used it for the SCID studies

as well.

This is what it looks like on its home page. You can see now it's serving the regional collaboratives. It's serving the state programs. And now, for those that have the little foot that looks like a DNA helix, those are the Translational Research Network components of the R4S site.

And I'll just go through quickly some -- this is just some screen shots of the SCID collaborative project, the various ways you can identify the different -- the many different forms of SCID that are available, that are out there. You can see that there's wide participation in the lysosomal storage disorder, as specimens and information begin to accrue, data display that lets you look at TREC results from the various laboratories that are participating.

Here you see some of the lysosomal storage disorders and the number of cases that have begun to come into that database. This is a shot from the SCID studies. You can see that in January to July of 2010, it was progressing fairly slowly. CDC had funded a

couple -- several states to begin doing screening.

NICHD came in and wanted to expand that much more rapidly and went out to California and New York, which have birth rates that really added to this database very rapidly. And you can see that, by January, April 2011, we were up in to the neighborhood of 14, 15 patients identified out of about 1.1 million babies who had been screened.

We're also in this long-term follow-up area now. We've developed those common information data sets that I alluded to that define the diagnosis data points and the interval data points that are used to monitor patients' response to treatment.

There are -- actually, because this is done at the point of care, there's a lot of demographic information, all the stuff you would do when you see a patient. And we're able to bring those in. It turns out that about 80 percent of the data points are common across all the conditions.

And we've already taken those to the national Library of Medicine for standardization and are working on the disease-specific kinds of

information around the conditions. And, as each new grantee comes in, that's one of the first things they do is begin to standardize their own languages for how they're going to describe things so we can move them back into the standardization system itself.

So I'm going to walk you quickly through just a broad overview, as the last slide. And, in fact, it's good that I'm able to see this. So, as you enter into the system, obviously, the newborn screening and the state labs are where newborn screening starts. They have the specimens. They have a contractual relationship with their population, who they screen.

As we move into short-term follow-up, the data about the diagnosis is coming back to the programs from the clinics that are involved. And that whole long-term follow-up process is beginning for everyone who has been diagnosed.

The Newborn Screening Translational Research Network comes in by providing that centralized data warehouse where we can capture the data from the multiple providers and investigators who are involved

in the studies. We bring in our different databases that relate to what the newborn screening laboratories are doing, the repositories that they have that become a research resource.

And then, as we move into our own infrastructure, we're using REDCap databases. It's a very commonly used database system now that evolved out of some work done at Vanderbilt. It's been taken up by 45 of the CTSAs, the Clinical Translational Science Awardee institutions, because we want to be aligned across multiple research infrastructures so that everything we do is compatible.

There's no personal health information in our databases. That is held locally, and we provide mechanisms to get that to local physicians who can relate back to the patient if any personal health information is required.

There's a whole series of back and forths that take place across all this stuff. The clinician and the researchers bringing data into the warehouses, the researchers who may ultimately want to access that data that's been collected for a prior study for a new

study that they think they can do, based on the data that already exists in these databases as we build on them over time.

And, on that, I will come back to the question that was asked of all of us, which is how do we relate to the Advisory Committee, or how might we relate back to the Advisory Committee. And I think, clearly, given the activity of the NBSTRN, it can facilitate the evidence development that can support nominations to the committee. That's only already beginning, though it's a bit ass-backwards at the moment, shall we say, in that the mandates often happen before we have the evidence coming into the databases.

Some day we may turn that around. But it includes the pilots of the new conditions, the clinical histories, interventions. But it does provide that resource for capturing post-market surveillance, which is common in orphan disease kinds of activities on the drug side of FDA.

They often will approve something early, based on their best sense of what the data says. But

they know they want to continue to monitor it to make sure they were right, over time. And by capturing this kind of data longitudinally, we have that post-market surveillance data component that may facilitate the committee's ability to look back and see how things are developing, presuming that the resources to maintain those groups and their data collection continues.

And every day we turn around, there's new bioethical and legal issues to deal with. And Jeff Botkin will talk about one of those. So, on that, I'll say thank you.

DR. HOWELL: Mike, thank you very much. The Translational Network, obviously, is off and running with lots of things happening and should be extremely profitable.

Questions or comments for Mike?

Ned?

DR. CALONGE: Hey, Mike, I think this is real exciting, exactly the kind of tool that will produce information useful to this group and clinicians. So both Terry -- although I don't see the

separation between the evidence world and the real world.

(Laughter.)

DR. CALONGE: I think this is a great interface that will inform both. So I'm not trying to be naïve, but one of the things that's come out of the genetic testing world is the inherent potential for misinformation out of something called GWAS studies, Genome-Wide Assessment Studies.

FEMALE SPEAKER: (Off-mike.)

DR. CALONGE: Sorry. I'm going to get there yet. And so, everyone knows GWAS. And it's a fascinating issue, because, you know, it's the old statistical rub; right? If you look for enough multiple comparisons, you'll find some statistically-significant results.

The other thing interesting about GWAS studies is even though you roll up all these small, increased risks, they don't account for very much in terms of actually additional predictability over other diseases. So the metabolic world's a little bit different; right?

One of the problems with GWAS studies is they never make the linkage of why the gene is linked; right? So there's no -- it doesn't require this scientific attachment of this gene creates this protein, which increase the risk through this mechanism. Metabolic conditions have a little closer linkage.

But I just have to ask the question. Are there opportunities for, kind of, those statistically-significant, but not clinically-important that impact potentially incorrect associations in looking across multiple metabolic markers in this method? And I'm thinking that the risk is lower, but I just want to make sure people continue to think that way.

DR. WATSON: No, I agree with you. You know, we're not ready -- for most things found in GWAS, they're not coming to newborn screening in, probably, in my lifetime. The difference is that, for these metabolic diseases -- the other things that we see in newborn screening, these are very powerful genetic factors. They're almost deterministic of disease.

What we have, then, to deal with is the variation across that disease that we learn about in newborn screening. And things like whole genome analysis are going to take us to the place where we could begin to interrogate the genome for those other genes that are altering the outcome of the patient with that very strong genetic factor. And I think, you know, that's going to be one of the areas of very interesting research that brings (inaudible) sequencing into newborn screening from a research perspective.

You know, as you move down into the weaker factors that are mostly what we find in GWAS, it's going to take a long time to aggregate enough of those to have actual utility and day-to-day care, let alone newborn screening. And I don't know that we're ready to go there in newborn screening. But, yeah, I think it's really that strength of the genetics that discriminates what one can look at in newborn screening and feel fairly comfortable that what you're seeing is close.

You may be biased until you really see the

general population and what's going on. But, yeah, I agree with you. That's his problem.

DR. HOWELL: Fred, do you have --

DR. LOREY: It's unfortunately already come to us. And it's causing quite a dilemma, because most of those grant applications require that that sequencing data be shared in DBGAP or whatever it's called.

DR. WATSON: DBGAP.

DR. LOREY: And be open to any -- yeah, DBGAP -- open to any other researcher, which violates our basic principles. So, in this first one, we reached a compromise where folks that are working with at Stanford simply wrote that in, that California would not agree to have this information stored. But, you know, they're not going to go along with that -- everything.

DR. WATSON: You know, I actually think the world's a-changing. You know, it used to be that when you thought about genetics research, it was this separate thing, you know, outside of practice. But, clearly, we're moving into -- certainly, in the

NBSTRN, in a point of care-kind of activity that's translational medicine.

Genetics -- as long as I've been in genetics, now, for 30 years, it's been translational medicine. We've learned from every new patient we see something else that informs us about the next patient we see. And these databases become very important, not just for learning, but also we're just beginning to think about how can a physician access this information to improve the way they care for their next patient, even if they aren't directly participating in collecting the data.

So DBGAP has been a problem. There's certainly been data limitation problems. You can't find Native American data in this database, because their own rules preclude their data going into DBGAP. So, as we move into what I think is where the health care system is moving, which is a learning health care system, that's the model we want to build the NBSTRN activities from so that we learn from our day-to-day care and variations in care, how to better care for the next patient that comes down the path.

And that's, I think, going to be a paradigm shift for NIH. And it's part of why they've been developing -- I guess, just yesterday, they funded NCATS, the Center for Translational Medicine at NIH. And, I imagine, they're going to have to start visiting some of these issues and thinking about how it differs from, sort of, what we thought about genetics research in the past.

DR. HOWELL: Thank you, Mike.

I think we probably really should go ahead. And I think it is worth commenting that we should commend the Eunice Kennedy Shriver National institute of Child Health and Human Development for putting a lot of money. This is a very expensive network. And I think it'll be extremely valuable to this committee and to newborn screening as a whole.

We're now going to hear from the Medical Director of ACMG. And Barry's going to talk about the regional genetics and newborn screening services across regional and national projects.

And one of the nice things about the Newborn Screening Translational Research Network, it can focus

on the research activities and build a structure using the regional collaboratives, which is funded through HRSA. And so, that's a very nice symbiotic relationship.

Barry?

DR. THOMPSON: Good morning. And a symbiotic relationship it is.

All of you know that the cooperative agreements that the Heritable Disorders program outlined and administered by HRSA allowed the NCC and the seven regional collaboratives to act on procedures developed and recommended by the Advisory Committee. And you're familiar with the seven regional collaboratives, I know. And the central goal of the regional collaboratives has always been to ensure that individuals had access to appropriate quality of care and genetic information and expertise in the context of a medical home.

And all of the activities of the National Coordinating Center work toward building bridges between the public health, primary care, genetics specialists, families, and the maternal child health

branch and facilitate the movement of quality genetic and NBS services to the communities and enhance the activities of the seven R.C.s by providing infrastructure coordination, technical assistance, and the resources that are necessary to eliminate some of the duplication of effort that has plagued us in the past. In the following slides, we're going to discuss a little bit about the NCC and its regional collaborative activities, both at the national and local level.

The initiatives include these seven items. And I'm just going to touch on each of those momentarily. The work groups are there to assist the regional collaborative efforts by doing such things as working with definitions, identifying and ensuring promising practices and engaging in activities that improve communication and linkages between the R.C.s.

I think everybody's familiar with the ACT sheets or the action sheets that have been developed and constantly under review and revision as clinical physician support tools for the primary care providers. The Evaluation Work Group is particularly

interested in measuring the progress made by the R.C.s toward the major goals and to identify areas of collaboration and technical assistance between the NCC, the R.C.s, and HRSA. And the emphasis is on finding commonly evaluative measures at that point that, not only give us a broad idea of what went on in the general issue, but in the specific regional collaboratives.

Long-term follow-up is exactly what it says it is. The joint effort with the NBSTRN's Clinical Centers Work Group to develop the minimum data set, particularly with emphasis on surveillance and public health measures to long-term follow-up and research. I need not say much about the medical home. That concept continues to evolve. And the idea is to bring some uniformity amongst the R.C.s in their definition and their applications for the medical home.

Publications Work Group coordinates the efforts between the R.C.s to articulate development to provide abstracts and session proposals, to increase participation, and reduce duplication of submissions to national meetings. The NCC's been particularly

interested in the successes that certain of the R.C.s have had in telemedicine and telegenetics and to develop an infrastructure in those R.C.s that do not fully employ that new technology on behalf of their patients. And there's a publication coming out of that work group shortly on telegenetics policy.

The interregional project on transition and opportunities for linkage with other centers and national partners works to increase uniformity in the approach of the transition model and facilitate the linkages between genetic expertise and the primary care provider. In most instances, you will recall that 80 percent of some of the pediatric providers talk about the importance of genetic information and the need for the application of genetic expertise to their patients. And the same proportion, talks about their inability to provide data in a cohesive and effective fashion for their patients and struggle with the implications that that has for quality medical care.

We're trying to move national-level issues to the local level by sharing information through a

variety of emerging topics. And, as Dr. Kemper mentioned in his presentation, one of those is certainly health reform and financing. Insurance, and, in particular, workforce development are key issues for us at the NCC and ACMG.

And the Coordinating Center collaborates with a variety of national centers outlined as on this slide below. These are important partners for us in bringing to the R.C.s through the NCC information that represents connectivity that the R.C. may not have with the national centers on their own.

We mentioned the ACT sheets as one of our educational and training programs and the genetics and medical home visiting professorships that have been a success. The idea here was to use funds from an NCC subcontract to sponsor genetic visiting professors and medical home visiting professorships, over the last two years, to provide an opportunity to enhance the medical home education for providers and families within an R.C. And there have been five of the genetics visiting professorships in the first year, a total of eight in two years, and five of the medical

home visiting professorships.

I need to acknowledge that the AAP, under a subcontract from NCC ACMG, has gone through a QIIN process for quality improvement integration network process, well-known to those folks who are pediatricians, that looks at the utility of the ACT sheets for pediatricians by soliciting feedback from a selected group of practices of all sizes and geographic distribution on the ACT sheet usefulness and utility.

I think everybody has seen the NCC collaborator. If you haven't, you'll hear from the editor, Judith Menkendorf. And she'll acquaint you with that, I'm certain.

Needless to day, it's a quarterly themed issue that showcases what's going on at the NCC and the R.C.s. Of particular importance to us, recently developed was the hearing loss brochure. It's a parent resource that highlights the importance of genetics as an aspect of hearing loss in the newborn period, particularly those patients that are screened as hearing loss positive at that point by newborn

screening.

In attempting to develop cultural competence to an increased degree, ACMG has sponsored two sessions at the last two annual meetings, the first on Native American perspectives involving the Native -- the Navajo Nation and the mountain states regional collaboratives work therein -- and then, the Vancouver one, Vancouver meeting, to look at CPT1A screening amongst first nations in the peoples of British Columbia and Alaska. It has two different approaches to the same sort of issue and the information provision to those populations in a way that addresses their cultural needs, perhaps different from the traditional patients that we deal with.

Long-term follow-up from the NCC has a variety of goals and a variety of deliverables that I won't go through as far as the short presentation is concerned today. But it's a bridge between the national centers funded by NIH and HRSA. And it's coordinating and accelerating long-term follow-up efforts by engaging in health informatic technology and standardization efforts and identifying the

intersection points between effective follow-up from our newborn screening grantees and other regional and national LTFU follow-up activities.

Again, mentioned earlier was emergency preparedness and the importance of the various aspects of needs of genetic patients when these natural disasters occur. Katrina being the example in medical home -- I'm sorry -- medical foods being the specific example of the difficulty of continuing to assure supply of critical medical foods to those patients who have been displaced by the natural disasters. And we've heard from -- I guess it was one of the previous speakers -- about the tabletop exercises that have been run in all of the R.C.s at this point using elements of the nationwide contingency plan under the Newborn Screening Saves Lives Act of 2007/8.

The educational activities and training activities are also important, particularly as cross-regional processes the genetics in your health brochures have allowed us to address specific needs at that point. And collaboration between groups such as the New York Mid-Atlantic Collaborative and the

Genetic Alliance Clearinghouse have been partnerships that have enhanced the NCC's efforts at education.

The annual metabolic nutrition and expanded newborn screening course is on dieticians and genetic counselors and genetics fellows to provide education and resources that will be important to those professionals. It was sponsored by the Southeast Regional Group. And also, the Sickle Cell Peer Educators' Training Program in the New York Mid-Atlantic Collaborative is one of those successful training programs that we'd like to highlight.

There are a variety of follow-up and treatment projects. And I'll only say a few words about each of those. The HIPPA-compliant registry of diseases under the IBEM-IS in region four is a priority program led by Sue Barry. And it's recently been shifted from HRSA to NICHD support with an award of a contract.

The EIF is a Web-based tool for sharing current information about a child's special health care needs involving family, specialists, and primary care providers a way to communicate during natural

disasters and other emergencies developed in region four with cross-regional participation and interest. The region one project that uses common data elements shared across long-term follow-up system with national and local partners and interregional participation has been going on since 1999. The Southeastern regional group has a specific requirement for long-term follow-up information systems and has been working with the development of a business plan requirements for that sort of activity.

We talked about access to medical foods, the nutrition management guidelines from the Mountain states is a consortium implemented to look for metabolic disease carefully and then share them both interregionally and nationally. And last but not least, the New England collaboratives quality assurance, quality improvement program, genetic systems assessment program, collaboration with Heartland, Mountain states and Western states, so a variety of activities moving on.

We heard about the region four project commenced in 2004. And it continues to expand and

currently involves, not only states from all seven of the regional collaboratives, but it's gone international with participants from several dozen countries. The goal is to improve quality laboratory, improve comparison and clinical validation of the tandem mass spec cutoff values. The program's headed up by Piero Ronaldo and currently called the R4 stork, or the R4S project at that point.

So regional collaboratives are feet on the ground, the people that are involved in the clinical and research laboratory -- research laboratory and clinical activities in a way that we aren't at the local level. But the Coordinating Center at ACMG allows us to draw those regional collaboratives together and to facilitate cross-development of projects, sharing of information, and implementation of projects that mean professional and personal success for those patients that need our help at that point.

DR. HOWELL: Barry, thank you very much.

Barry is going to be around. And I think if you have any comments or questions, please try to nab

Barry later, since we're running a bit behind time. And I'd like to move along.

And we'll hear from Carla Cuthbert, who is going to discuss the laboratory quality program. And Carla, as most folks around the table know, is responsible for the CDC's newborn screening molecular biology branch.

Carla?

DR. CUTHBERT: Thank you. I'm Carla Cuthbert. And I'm here to talk to you about the quality -- the laboratory quality program that has been present at the CDC before coming on -- a little over 30 years now. And I'm actually going to be talking to you about the role of the branch of which I'm Chief, the Newborn Screening and Molecular Biology Branch.

Now, CDC, acting through our branch, has been given a mandate by Congress, through the Newborn Screening Saves Lives Act that we've been hearing about a lot. And we have been asked to provide for quality assurance for laboratories involved in screening of newborns and children. And we provide

quality assurance for newborn screening tests, performance, evaluation services, technical assistance, technology transfer. And we provide appropriate quality control materials to evaluate performance of new screening tools.

And the approach that we're actually using to do this is through a series of teams that we actually have in our branch. And I'd like to let you know that we actually have six teams. But the four teams that are most relevant and that interact with the public health laboratory system the most are the ones that are indicated here.

Most people will be able to identify or have heard about NSQAP, which is the Newborn Screening Quality Assurance Program. And that, again, has been in operation for a very long time. And what we also do have is three other teams called the Newborn Screening Translation Research Initiative, or the NSTRI. And I'll be describing these teams and their activities in a little bit more detail.

And two new teams that I recently developed in the last few months, actually, were designed to

specifically address many of the specific technical issues associated with newborn screening. And that's the biochemical mass spectrometry laboratory and the more recent molecular quality improvement program. And again, that's in direct response to what has been happening as a result of the Advisory Committee and as a result of what we're actually seeing as gaps within the public health -- laboratory public health system.

So I'm going to talk about the first team, which is the newborn screening quality assurance program, which many of you already know to be the only comprehensive quality assurance program using dried blood spots for newborn screening. And we provide a number of different activities and services to the newborn screening laboratory community, which includes filter paper evaluation for new lots of filter paper.

We provide reference and control materials. We provide a system for efficiency testing. We have on-site, online Internet reporting for the laboratories. And we have a very strong program of following up of any false/negative results.

We have special -- we have specific subject

matter experts to special scientists within the branch that will follow-up on any of these cases with the states, with any laboratories to make sure that, you know, it's not just a clerical error. If there are any issues associated with any technical issues, we try to address those very appropriately.

We also play a very important -- well, we also have a very strong desire to have a lot of training, consultation, and network resources. Many of the activities that we do provide are coordinated through our cooperative agreement with the Association of Public Health Laboratories. They are a very, very close partner, and rarely a day goes by without my actually interacting with them in one way or another.

With respect to some of the things that have happened over the course of 2010 -- and again, these are just statistics, but will just give you a sense of our activities throughout the year. We have 100 percent participation in the newborn screening laboratories that are involved in screening in the United States. And again, this is a voluntary process. And all of the states are very, very willing

to participate with us. And we have very good relationships with them.

We are also able to expand some of our activities to 67 countries. And again, this is voluntary for them as well. Last year, over 700 dried blood spots were actually produced by our scientists within the laboratories.

We had 20 employees that are involved in this particular process. And that's shifted a little bit, because we're now incorporating molecular into this particular program. So we have a very vibrant group of scientists who are actually involved in the process of providing quality materials to the states.

In terms of new enrollment, these are laboratories that have requested to participate in our program. And at the end of last year, we had over 460 labs enrolled. We do have a laboratory -- the one thing that we require of our laboratories, of course, is that they send in data. And you'll find that the numbers that I have here, in terms of the numbers of labs participating in either proficiency testing or quality control or any of our programs, they're

required to submit data. And when they don't for an entire year, we do drop them, because there is a waiting list, in many cases, for specific programs.

This just gives you an idea of the 67 countries that are participating in our quality assurance program. You'll notice that there's a distinct absence of the decrease of participation in Africa.

We do have a wonderful collaboration that we are engaging in with the country of Ghana. And Ghana is actually one of the first countries that is really moving towards nationwide newborn screening. This is for sickle cell.

And we have a wonderful collaboration that I will mention to you very briefly that will also support our program here. The NSQAP in a program provides quality assurance materials in dried blood spots for a number of different conditions. And these are all listed here.

One of the ones that we have most recently been providing support for is the combined immune deficiency. And we are very happy to have a number of

different states participating in that program.

So the second team that I want to just bring your -- draw your attention to is the Newborn Screening Translation Research Initiative. It's a smaller team that represents an ongoing collaboration between the CDC Foundation and our branch. The mission is to assure the translation of research methods into routine laboratory tests for newborn screening and to ensure that it leads to sustainable, high-quality testing.

The team itself develops newborn screening methods. And again, we need to have methods in operation within our laboratories so that we can actually provide support -- technical support -- for the labs as we bring them on. We interact with the state public health laboratories in the translational process.

And we are very much interested in adapting various innovative technologies for screening and quality assurance. And we work very closely with the newborn screening laboratories, again.

There are a couple of ongoing laboratory

projects in this particular team. One of the most important, the highest priority for them is severe combined immunodeficiency. And they have spent some considerable time being able to produce various proficiency testing materials for the TREC assay.

And TREC stands for the T-cell Receptor Excision Circle assay. And that's the assay that is predominantly being used for SCID testing or for SCID screening. They have a method that has been developed and we've been very actively engaged in providing training for personnel and providing various forms of technical support for the laboratory personnel as they implement and bring on this particular test.

There is also involvement in lysosomal storage disorders. And again, we provide Q.C. and P.T. materials for these five disorders named here. And again, we also provide training for personnel and technical support.

The third team that I want to bring your attention to is, of course, the biochemical mass spectrometry laboratory, which has recently developed and has a mission of working with public health

partners to develop new mass spectrometry-based assays to detect and monitor metabolic disorders and to enhance newborn screening laboratory performance through innovative approaches. Two of their highest priorities are to develop new methods using this technology and to develop other pilot programs looking at tandem mass spectrometry analytic ratios as part of their proficiency testing endeavors.

In terms of public health impact, there is 100 percent coverage right now of the primary biomarkers for the 43 disorders. They have Q.C. programs, and they work together with the previous team for the lysosomal storage disorders, because there are tests that are based on mass spectrometry for that particular -- for lysosomal storage. And again, they provide Q.A. materials to enhance analytical specificity through second-tier testing.

The molecular quality improvement program is one that is of high priority to the branch itself. And this particular program was developed as a result of, again, the recommendation that the Advisory Committee had last January when they recommended SCID

through the panel and again, when Secretary Sebelius accepted it in May last year. So we've just definitely recognize the need to provide support for the public health laboratories as they worked towards bringing molecular testing into their routine practices.

So we're looking at either what the second tier primary molecular methods that are being integrated. And again, molecular screening, again, brings a very different and a new technology into the newborn screening laboratory. And we need to make sure that best practices are being developed.

This slide just indicates that, at the end of 2010, 36 states, that are shown in green here, have been offering a molecular test. And again, this was not state-wide, necessarily. This would have been with targeted populations. So, as you can see, these states are now looking at what the incorporation of SCID, looking at doing state-wide testing and testing all of their population.

So in terms of activities of this particular group, they have played a very -- they are in the

process of establishing what's called the Newborn Screening Molecular Network. And again, that's this little icon on the right here, that brings together APHL, the public health laboratories, and our branch together to share common knowledge and to identify gaps.

We have established and implemented a molecular assessment program, which is really just a site visit that allows us to visit different laboratories and take a look at how they're doing with their molecular implementation. This is already in progress.

We've had two visits so far. And we're having a third one before the end of the year. And again, we're just looking at identifying best practices and making sure that all of the laboratories are well-equipped with being able to perform this kind of testing.

We are, of course, providing quality assurance research for the development of materials, because, again, it's a very different process from using -- from developing materials for, say, the mass

spectrometry or the inborn errors in metabolism conditions. Here, you actually have to have the appropriate mutations, and everything else has to be, quote, unquote, "normal." So we do have to provide appropriate materials.

Molecular characterization has to be very well-done. And we also have other translational research projects that are involved.

There are three main priorities at the branch. And again, these are to -- primarily, the first one is to sustain and strengthen our existing quality assurance programs.

The two main conditions that we are focusing on here are cystic fibrosis DNA. And we are working with California to be able to improve the number of samples and the number of -- the variation of samples that we actually have. So that's something that we're very excited about.

And again, I referred to our collaboration with Ghana. You'll notice here in this table below that Ghana, while it has a population of about 24 million, it has about 13,000 sickle cell disease

births every year. And this is in comparison with the United States with about 308 million with just barely 2,000 sickle cell births each year.

So we have been engaged in a collaboration with the Ministry of Health in Ghana, the hospital, and laboratory. And again, this is work that has been initiated by a previous member here, Dr. Kwaku Ohene-Frempong, who is -- of course, you know, he's a wonderful human being.

And we're so delighted to have been able to make these connections. And I think he's currently in Ghana right now. And we are actually working at making this go.

They are going to be able to provide samples for us so that we can actually use them in our program. And in return, we're going to be able to provide technical assistance and bring them into our sickle cell program. Again, they are the first African country to want to do this nationwide. So that's a very good plus.

Our second main priority is to, of course, implement quality assurance programs for any recent

additions or any new additions to the newborn screening panel as per the Advisory Committee. And the most recent one was SCID. So, as was mentioned earlier by Mike, we have been able to support Wisconsin and Massachusetts and the Navajo population for a few years with some funding for SCID implementation in newborn screening.

And, as of the next week or two, we will be able to fund another two states. And they've not been announced. I would be happy to share them with you, but I'm going to have to wait another week while we get all of our paperwork done. But we're very excited about those two new states that will be joining and getting funding from us.

Of course, we have an ongoing proficiency testing program that is moving from the pilot phase into the routine activity of NSQAP. And that right now is underway. And currently, we have a little over 11 participants. And, of course, we have that method, a method that we've already developed.

And then, finally, our third major priority, of course, is to identify gaps, specifically with

respect to newborn screening implementation regarding molecular testing. We've already established the MQIP, or the Molecular Quality Improvement Program. The network, again, involves all of the newborn screening laboratory persons within the United States. We have already initiated the molecular assessment program. And we are going to be presenting some of the initial outcomes at the San Diego APHL meeting in November. And again, we're involved in collaborative research studies to make sure that we are able to assure molecular testing.

So that gives you the highlights of what we're actually doing. And this just gives an indication of our team leads and a very dedicated staff that we have at the CDC involved in this project.

And thank you, again, so much. We are so very happy to be a part of this particular team. No one ever wants to be alone when they're working. And it's a very different relationship that we have with our newborn screening community that's not always evident in our laboratory division.

So thank you. And if there are any questions, you can find me somewhere outside.

DR. HOWELL: Carla, thank you very much. Your program continues to be the world leader, obviously, in quality assurance. And everywhere you go, you find there's a lab that's a member of your Q.A. team. So thank you very much. And we're glad that you're continuing to collaborate with Kwak in his programs in Ghana.

The Newborn Screening Saves Lives Act did not have any legislation tied to the military. But there have been some really important changes in newborn screening in the military, which Mary Willis will discuss with us next.

Mary?

DR. WILLIS: Okay. Well, I'll try to go through this quickly. I'm a clinical geneticist. I work for the Navy. And I am also the representative for the DOD on this committee. And today, I'm going to be talking about newborn screening for the military dependents.

A lot of people may not know that there's

anything different about military babies. But hopefully, I'll highlight what's going on. I'll just go over a little bit of a history and then talk a decent amount about the new contract that's been established with Perkin Elmer Genetics.

So some facts about military babies: There's about 120,000 babies born to military families every year. That's about the same as is born in, say, Michigan. Half of those babies are born at what we call MTF. And this is the military, so you have to get used to these three-letter designations as things.

MTF are bound by federal law, which trumps state law. And so, they are not obligated to use state lab systems or report their positives for newborn screening to the state health departments. However, many MTF do choose to comply or attempt to comply with state law.

Military individuals, as most people understand, are a very mobile population, but more so even than I realized until I worked for the military. So patients and families are not just moving around because they're being stationed to new places. But

with deployments, a lot of times, families will move home while their active duty member is deployed. And sometimes, that's within a couple days of birth.

Also, physicians, if they're active duty, are a very mobile population. So the person you used to be able to call and ask questions is not necessarily the same person as that physician. And, of course, the military is worldwide, not just in the United States.

So a little bit more about the MTF: There are 93 MTFs worldwide. And 52 of these are doing deliveries. An additional 21 are involved in newborn care. And so, they may be sending newborn screening, especially if the babies are born in a foreign country and then come up for their newborn -- you know, newborn visit to these MTFs.

These are located in 31 states and 10 foreign countries, which I have listed there. Births -- and again, here's an acronym. CONUS stands for Continental United States. And OCONUS is Outside the Continental United States.

(Laughter.)

DR. WILLIS: So CONUS is about 62,000 births, and OCONUS, about 6,500 per year. The largest volume is Portsmouth. And that's 290 babies a month. That's a lot of babies at a single hospital. And the least would be Guantanamo Bay in Cuba. And they get about a baby a month.

So some background about newborn screening: The first, sort of, official thing that went on was in the Army. And that was a policy was published requiring MTFs to screen for at least four disorders. That was in 2002. And to also have a written policy and procedure in place to do newborn screening.

As everybody in this room knows, the big thing happened in 2004. And that was approving the report by the ACMG for universal screening of this panel.

Well, two months later, the AAP and the March of Dimes endorsed the panel. And this is very important for the military, because -- I've got a quote there from the TRICARE manual. The TRICARE manual is what dictates what we offer our dependents and our patients.

And it says that we will do the screening in accordance with the American Academy of Pediatric guidelines. So it wasn't until the AAP said, yes, we think this is a good idea that we really needed to move forward.

But as soon as this went forward, people in the military starting to say, hey, wait a minute. Some people in our -- some of our dependents are not getting equal benefits, depending on where they're being born, if they're sending to the -- newborn screening to the state, what's going on. And they started adding up the total number of babies we might be missing.

And it was a significant number of babies. And so, things started really moving at that point.

The Navy was the first to act. They have a group called the Perinatal Advisory Board. And that is a group of perinatologists, neonatologists, pediatricians, and O.B. doctors and nurses. And they decided that this was something we needed to do and we needed to do now. And they asked the Navy lab community to figure out how are we going to do this

expanded screen.

And, in November, the Navy lab people came back, and they said, "You know what? We can do some contracting. We can find out a way to get a single laboratory to do all of our testing for us." And so, in the Navy MTFs, they started doing universal screening through what was then pediatrics, which has now become Perkin Elmer Genetics, for this expanded screening.

TMA, again, an acronym, initiated a cost estimate. What was it going to cost? What if we did this DOD-wide? What if we had a single contract that we could offer to all of our MTFs to do all of these disorders?

And so, we have to figure out, well, how much is that going to cost us, and is that going to be a good idea. And it was informally endorsed that that was a good idea. So again, things can move forward.

The IPT, Integrated Process Team, was formed to facilitate military health service-wide implementation of newborn screening. That was in 2005. And again, that's a time when there was a lot

of disparity between different states and what they were offering as opposed to now, when most states are doing about the same screening.

Health administration policy recommendation came out. And this was the three tasks for the military IPT: education plan, a newborn registry, and a centralized contract. And I'll go through each of them.

So the IPT, over two years or so, developed a curriculum targeted at provider groups who were going to be involved in the newborn screening care. And the authors -- the primary authors of that were Scott McLean, who was my predecessor on this committee, and Katherine Camp, who's frequently at these meetings. But I haven't seen her yet today.

And they came up with this curriculum. We also borrowed some educational tools and designed some for ancillary staff and for the parents. And then, once these tools were available, we basically handed them back over to the different services -- Navy, Army, Air Force -- and said, "Okay, now, use this. Educate your people."

And they were made available on a Web page. And this is a simple Web page, if you go to it. It's not sampy. It has mostly just links to other things.

The education plan is there. A PowerPoint is there, some things we borrowed from the AAP as far as the brochures. And I would like to put, me, personally -- this is not me, the DOD. This is me, the geneticist -- would like to put links to the Baby's First Test Web page on there as well.

The registry -- when this was initially thought we were going to have a single place that was going to do screening for all military babies, we thought, well, then the registry needs to be able to talk to the people providing this data. And so, work on the registry was put on hold until we knew who the contractor was going to be for that testing.

And now that we have that contractor, things are moving forward on the registry. I'm not quite sure how this is going to look. It's very early in the process, but it's going to be similar to the way that we direct mammograms and colonoscopies.

Now, I won't go through all of this. But

basically, the solicitation is what we asked for in the contract. And so, that's important, because when you ask for things in a contract, and then, you get those things. And if you didn't ask for something in the solicitation, then it's not necessarily part of the contract.

But some issues -- of course, we wanted the (inaudible) test. We wanted daily, secure, worldwide, electronic reporting, because we have a worldwide population, consultative services five days a week, because that seemed to be what was going on around the country. We wanted it to include screening materials, et cetera. And then, of course, we wanted it to link to this potential registry.

So what happened with the contract -- the pre-solicitation notice was placed on FedBizOPPS, or Federal Business Opportunity. It was actually first put there in '07, but then, there was a lack of activity for a couple of years, couldn't get things rolling. And so, it was placed back on FedBizOPPS in 2009.

And then, the actual solicitation was put on

the Internet bid board system for the Defense Logistics Agency in May of '09. And the contract was finally awarded to Perkin Elmer Genetics at the beginning of this year, in January. The contract went into effect May of this year.

And then, the action memo, which is basically our marching orders, was signed July 1st. And some details about what that action memo is -- that comes from the Assistant Secretary of Defense for Health Affairs, Jonathan Woodson. And the contents of that action memo -- there was a lot of background information: Why is newborn screening a good idea? Why did we start this process? Contract modification can be done -- or disorders that are recommended by the AAP -- you'll notice not this committee, but the AAP.

And I think, in response to the fact that, when the process started, there was a lot of discrepancy in what disorders were being screened, but now, not so much, instead of making it a universal mandate -- everybody has to use this contract -- basically, what it says is we encourage you to use

this contract. But we are also asking you to evaluate what you're currently doing and then make the right choice, clinically and economically, for your MTF, or, actually, for your service. And then, that trickles down to the MTF.

Some details about the contract -- it's a five-year contract. Contract pricing -- I debated whether or not to tell you the price. But it's public knowledge, so there it is: $33 per baby for CONUS and $32 -- I'm sorry, $33 per baby, CONUS, and $32, OCONUS.

And for the OCONUS -- these are two separate contracts, actually. OCONUS does not include the shipping of the samples, because, depending on where you're shipping from, there can be a lot of complexities. And so, they decided to leave that up to the MTFs to get their samples in.

There is some very specific things about receipt of specimens and satisfactory specimens and when we have to hear about those, results reporting, three-day turnaround, HIPPA-compliant. We were pretty specific about what we wanted their reports to tell us

as far as the disorders screened, et cetera.

Rescreening and confirmatory testing -- so if the laboratory says we need another blood spot on this baby, that is actually -- you know, for confirmatory testing, for an unsatisfactory sample, that is under the same $33 cost.

For abnormal results, we actually wanted to know the number. What was your tyracine, not just that it was abnormal, which has not always been part of the reporting that Perkin Elmer has done.

We wanted detailed interpretation of what those results meant and recommendations for additional testing or confirmatory studies. And we wanted a contact person that the pediatrician could call if they have questions.

Part of the contract is that Perkin Elmer will report this data to the states and to the Genetics Resource Center, if we so choose. And so, as an MTF signs up under this contract, then, Perkin Elmer is supposed to contact the state where that MTF exists and say, "Okay, now we have some data for you. How do you want it"? I'm not sure if that's actually

happening or how it's happening, but that is part of the contract.

The consultative services -- again, what we asked for and what we got -- genetic counseling 24/7. And these consultative services will include interpretation of the results, recommendations for evaluation for their management, educational support, and patient referral management. And that's, sort of, broad. And we're trying to figure out how that should look.

We wanted Perkin Elmer to -- they're the person who's contacting the pediatrician. They wanted the pediatrician to know what to do. And I'll talk to you about, well, what do you do with these positive babies in the military, since we don't have a military newborn screening program. This is a test.

There is an issue about training and education that says, basically, how do you do a blood spot and how do you make them good spots so you don't have to be rescreening babies. And they have a quality assurance thing in place where they'll look. And if there's a certain MTF that's sending a lot of

unsatisfactory specimens, they'll go back out and reeducate to make sure that we don't have to keep doing those rescreens.

So prior to the contract, this is the list of MTFs -- and I'm sorry about the small print -- that we're using Perkin Elmer Genetics. And if you think about the history, it, sort of, makes sense. Most of these are Navy, because Navy started this a while ago.

There are a number that are in the OCONUS locations, because, again, that makes sense. They needed to get -- they wanted to get American, if you will, newborn screening done on their babies. Or, say, down at the bottom, offered in Nebraska -- Nebraska is testing labs. It's actually Perkin Elmer Genetics. So they were already going there.

This is the list of MTFs that are utilizing the contract. This is a shorter list than the previous list, obviously. And that has something to do with an old contract needing to run out, some technical points. But we anticipate most of those on the previous list, which will become part of this list.

There are some MTFs that are new to this list. Korea is now sending theirs. Interesting -- one of the few Navy hospitals that wasn't using Perkin Elmer before in Pensacola before is now using Perkin Elmer -- and Brooke Army Medical Center in Texas.

So, as far as interactions with the state programs -- and there's -- I've gotten a lot of questions just one-on-one about this sort of thing. What about the difference between state law and the tests that are being done by Perkin Elmer?

Well, each MTF must decide what they're going to do about that. So if there is a second screen, which is part of either law -- for instance, in Texas -- or highly recommended, as it is in Maryland, that MTF has to decide, well, are we going to try to do that second screen. Bethesda currently does not do a second screen.

Perkin Elmer will charge that $32 or that $33 again for the second screen. But they don't treat it as a second first-time screen. They do track the babies and say this is a second screen. And that's how the data would be reported to the state.

As far as additional disorders, for instance, New York with SCID and Krabbe, Keller is not part of the contract yet. But if they were to become part of the contract, they would have to decide what to do. Perkin Elmer will do SCID testing, which they already do, for an additional fee above the cost on the contract. And they will do it on the same blood spot card.

Krabbe they don't do. And so, that's not an option. And I don't know the -- Keller would have to figure out what they wanted to do about that.

Since reporting the public health data is part of the contract, we need to make sure that that is happening. And we need to keep going back to them and talk with the states and say, "You know, how do you want this data," and also talk to Genetics Research Center and say, "You know, how do we want this data? And is this useful data"?

But it's the public health data that's being reported to the states and not the individual positives. And that has been a source of confusion, actually, for some of the military physicians.

They're just assuming, "Okay, I've got a positive screen. The state's going to take over, and it's going to be fine." But that's not the case.

The state programs should not be being asked to do follow-up for the positive screens at Perkin Elmer. What needs to happen is that baby is referred for appropriate follow-up. And, in many cases, the doctors doing that follow-up will be the same as the doctors doing follow-up for the state programs. But it needs to go through the right channels. It needs to go through our purchase care network to those physicians.

So each MTF, again, is going to have to figure out their referral pattern. And these referral patterns are, in many cases, already in place. It's going to depend on what the disorder is and where that baby was born. So the OCONUS locations are going to have to figure out, is this a baby that needs to be transferred back to the United States or not.

It's a big deal to transfer a baby. It's a big, expensive deal to transfer a baby and their family back to the United States. So, for instance,

if a baby is born in Cuba, and they have phenyl hyperthyroidism, that's actually treatable. And they can stay.

And military pediatricians are used to taking care of kids, with help over the phone. And so, that is what has happened, is those babies have stayed where they are. However, proprionic acidemia -- most likely, that baby is going to need to be transferred.

As far as who's going to do the follow-up, well, military physicians -- there are a number of them that could take care of cystic fibrosis, hematologic disorders, or endocrine disorders. But they're at the big centers like San Diego and Bethesda. And so, depending, does it make sense to move a family so that they can get care at one of those centers, or should we refer to our civilian counterparts that are in the area.

For the metabolic diseases, truly, there are very few metabolically-trained clinical geneticists that work for the military. I'm one of the very few, which is probably why I have this job. And so, we are

going to have to be referring the vast majority of those babies out. And again, those are going to be the same physicians that are doing the follow-up for the state programs, but the way that they get there is a little bit different.

As far as additions to the panel, the obvious question is what about SCID. And I will remind you about the TRICARE manual, which says that we need the AAP to endorse SCID. And so far, that has not happened.

And so, until the AAP does something official to endorse the addition of SCID to the panel, we can't renegotiate the contract. So we're, sort of, waiting for the AAP to do that. Now, AAP already acted on congenital heart disease.

DR. HOWELL: Yes.

DR. WILLIS: So we're hoping that they're going to come up with something on SCID soon so that we can renegotiate the contract to add that. And I think that's all I have.

DR. HOWELL: Mary, thank you very much. That was an extremely informative thing. I have a

slide that talks about how cheap newborn screening is. And I compare it to what we spend on Lipitor.

(Laughter.)

DR. HOWELL: And using the figures that you just presented, newborn screening in this country, if we screened everybody for what you're paying, would cost one-half week expense of Lipitor in this country. So that gives you an idea of how cheap it is. That's why I don't like to talk about cost of newborn screening, because it's such a bargain.

We've run considerably over time, but we had a tremendous lot of really great information, which we appreciate, from the various and sundry group. And everybody stayed right on schedule. But what we're going to do is we're going to return later, because the folks in the audience, in particular, need a fair amount of time to get a bite to eat. But we're going to start again at a quarter of two. And we'll start right on the minute at a quarter of two. Okay? 1:45. Thank you.

(Break.)

DR. HOWELL: We're going to have Seth

Morris. Seth is here with his parents. And Seth is going to -- Seth himself has phenyl ketonuria. And he has a brother who died of Krabbe.

And, Seth, I'm going to ask you to -- you can come up here with your dad, and you can sit down at this microphone and comment. And you can bring your dad or your mother or both or whoever you'd like to come along. But we're looking forward to hearing from you.

Seth's birthday is on June 14th, which I told him is a very good day. It just missed my birthday by a few days, which is very good. Being a June baby is an excellent way to start.

Okay, Seth, are you ready to roll?

MR. MORRIS: Yeah.

DR. HOWELL: Let's roll.

MR. MORRIS: My name is Seth Morris, and I have PKU. PKU is a disorder that makes me unable to process certain proteins like meat and beans. Luckily, I was diagnosed at 11 days old and treated. Untreated, I would not be the young man you see before you today. I'm a cornerback on my school's football.

I'm a catcher on the baseball field. I am an A student, and I'm a big brother.

I wish my little brother, Grayson, could have had the same chance to be what I have become. Grayson had Krabbe Disease and died six days before his first birthday. Texas does not screen for Krabbe like they do PKU.

Why is my disease so much more important than my brother's? Why should his life be any more important than mine? Why me?

This summer, I saw Krabbe kids for the first time, kids that were screened for and treated. They are running and laughing and playing. But my brother didn't get that chance. He never even crawled.

Everyone should get a chance at life. My life should be no more important than Gray's. I will have to live with that thought every day for the rest of my life. But you have the power to change that. Please help me make a difference. Thank you.

DR. HOWELL: Thank you very much.

(Applause.)

DR. HOWELL: Thank you very much, Seth. And

your presentation, as you know, will go into the record of this committee. And you'll be able to see what you had to say. But that was excellent. And I think that you're a tremendous testimony to the effectiveness of early diagnosis and treatment of phenyl ketonuria. And we appreciate that.

Does anybody have a question of Seth? He obviously has a great deal of wisdom there.

Thank you very much, Seth. And we will look forward to following your career. How is your team doing, your football team?

MR. MORRIS: Good.

(Laughter.)

DR. HOWELL: It better be, since you're the quarterback; right?

MR. MORRIS: No, I'm the corner, not --

DR. HOWELL: Oh, I'm sorry. Okay. All right. Good. But anyway, I'm sure you're a pillar of that outfit.

MR. MORRIS: I'm missing a game today.

(Laughter.)

DR. HOWELL: Oh, goodness. Do you need us

to write you an excuse to take to your coach?

(Laughter.)

MR. MORRIS: No, sir.

DR. HOWELL: We'll be glad to write you a note and say you were doing worthwhile things, and so forth, et cetera. Okay.

MR. MORRIS: Yeah. I just hope my Q.B. doesn't get hurt, because he's the only Q.B. that we have for my team.

DR. HOWELL: Oh.

MR. MORRIS: Each team only has one Q.B. So --

DR. HOWELL: Okay. Great. Thanks very much. Great job.

MR. MORRIS: Thank you.

DR. HOWELL: Super.

(Applause.)

DR. HOWELL: And we're going to go next to Sharon Terry.

And, Sharon, you've been around a long time, but seldom have you had an act so hard to follow.

MS. TERRY: Yeah, absolutely. And I'm also

aware that we're about a half-an-hour behind, so I'm going to cut a half-an-hour out of my comments.

(Laughter.)

MS. TERRY: I want to thank you, Dr. Howell and members of the Advisory Committee. It's my pleasure to provide comments today on behalf of Genetic Alliance and Baby's First Test.

During the past seven years, this committee has made very significant and a lasting impact on the welfare of newborns and children across this country. And here is where, really, I did write all the accomplishments. And I'm going to skip them all, since we have heard today about how wonderful the committee has been.

DR. HOWELL: But they'll go into the record.

MS. TERRY: Yes. I will.

DR. HOWELL: Okay, good.

MS. TERRY: These advances have enjoyed your exceptional leadership, Rod. Your passion, your drive, and your wry wit has driven this ambitious agenda. You have a grace that allows you to navigate the rapids with aplomb and also still face the hard

questions.

Thank you for guiding the committee for all these years. I have witnessed the urgency with which you have led the committee to grapple with emerging topics and create frameworks to better strengthen and support state newborn screening programs.

Due to the solid foundation developed during the past seven years, this committee is poised to address the emerging issues facing the entire spectrum of population-based screening, including whole genome sequencing, the public trust, incidental findings, and much more. Even as technology advances and new priorities emerge, the leadership of this committee has an interest in children and their families central to decisions and recommendations. As a mother of two children diagnosed with a rare condition, I appreciate that piece above all.

To Dr. Howell and to the other departing members of the committee who are rotating off this year, the advocacy community and the 4.2 million babies born each year, thank you for your vision and your commitment. Thank you.

(Applause.)

DR. HOWELL: Sharon, thank you for your kind remarks.

We now have Katherine Harris, who's going to talk about NYMAC.

And here comes -- Katherine, why don't you come up and sit at the front, rather than the microphone back there?

We had a very nice note from Katherine's associate, Michelle Caggana, who is not able to be here.

MS. HARRIS: So she tasks with me this welcome. NYMAC welcomes this opportunity to thank Dr. Howell for his longstanding support of programs serving people with special health care needs.

Under your leadership, the Secretary's Advisory Committee has set standards for newborn screening never before thought possible. Finally, in this national forum, newborns, regardless of the state in which they are born, have the same chance to be diagnosed with so many devastating conditions and receive the treatment they need to live healthy and

productive lives.

The members of this committee and its subcommittees have engaged in thoughtful and intelligent discussions around guidelines and availability of screening, medical care, and treatment that are bettering the lives of so many. I personally am grateful to have worked with Dr. Howell for over 20 years, first, through the regional networks and now the regional collaboratives, to bring to the national stage the issues of uniformity of screening and evidence-based care.

I also am grateful that Dr. Howell was able to participate in last spring's NYMAC summit, bringing his insight and wisdom to many people who had not yet heard of his work. As a project manager of NYMAC and personally, I want to wish Dr. Howell well as he steps away from this committee. I hope that he leaves knowing that it will continue doing well the job he has set before it.

DR. HOWELL: Thank you, Katherine.

(Applause.)

DR. HOWELL: And, obviously, all those kind

words go to all the other hard workers that are rotating off this committee.

Next, we have Jennifer Garcia. I do not see her.

So we'll move on to Christine Brown from the National PKU Alliance.

MS. BROWN: Thank you. My name is Christine Brown. I'm the mother of two children with PKU as well as the Executive Director of the National PKU Alliance. I would like to thank Dr. Howell and the committee for your leadership and vision in making sure that the voices of children and adults with heritable disorders are heard.

As we all know, PKU is one of the most prevalent diseases among the heritable disorders, but the National PKU Alliance is still a newcomer to the national rare disease space. And we are still learning to navigate federal policy and the players involved and the guidance and the insight. And the relationships that Dr. Howell and others on the committee have helped me to foster have been really integral and critical to our success and our work.

I simply do not know where I would have turned, without having this committee in place. And your work, in particular, the work on medical foods, and the issues around access and reimbursement of medical foods, has been paramount in our success in order to bring that to the attention of both state and federal legislators. And, as Alex alluded to earlier today, that fight is not over.

Right now, we're currently waiting for the essential health benefits package to come out of HHS. We hope that will happen by the end of the year. If medical foods are not included as essential health benefits, that essentially means that states that still want to cover, or have insurance cover, medical foods are going to have to do so at their own expense. And so, that possibly could put about 34 current state laws in jeopardy.

So I'd like to thank you for making a difference in the lives of the 15,000 Americans living with PKU in this country.

Thank you, Dr. Howell, very much for your leadership and support and insight. We hope that the

committee will continue to welcome and count upon the voices of children and adults in this country living with heritable diseases. Thank you.

(Applause.)

DR. HOWELL: Thank you. And I'm sure that the committee will continue to be interested in medical foods and will pursue whatever opportunities come up there, and so forth.

We have next Dr. Celia Kaye representing the Mountain States Genetics Regional Collaborative. I know she's --

FEMALE SPEAKER: She's not back from lunch yet.

DR. HOWELL: She's not back from lunch yet.

Jill Levy-Fisch is back from lunch. I've seen her. And she's on the next -- and Jill is Executive Director of Save the Babies Through Screening Foundation.

Jill, why don't you come up here so we can hear your mellifluous tones better?

MS. LEVY-FISCH: Thank you for the introduction. My name is Jill Fisch. I am the

president of the Save Babies Through Screening Foundation. We are the only advocacy group in the country solely dedicated to newborn screening.

In honor of Newborn Screening Awareness Month, we have launched a redesigned Web site and an educational video entitled, "One Foot at a Time." Our user-friendly site provides quick references for people in various circumstances: practitioners, expectant families, families whose baby has had an initial positive screen, and families whose child has a confirmed diagnosis. There will be an interactive area where experiences and information can be shared.

We also include an FAQ section regarding newborn blood spots. The information for both the Web site and the video was developed by our network of parents with firsthand experiences of newborn screening supported by the knowledge of a medical advisory panel with vast combined experiences in newborn screening as well.

In order to help parents become more informed, we developed the educational video to give families a new way to learn about why testing is

recommended, when and where it will be done, how to obtain results, and how the process can be more comfortable for parent and child. The video was designed for use during pregnancy or even before, where parents can learn in a more relaxed setting.

It can be viewed on our Web site, (inaudible) YouTube. DVDs are available at no charge. And we also have a Spanish version. We're pleased to announce at this time that we have signed an exclusive licensing agreement with the state of California for the use of the video, which makes California a true leader in newborn screening education.

One of our advisors on the video was Dr. Howell.

Dr. Howell, you wove together a successful collaborative effort after your appointment to this landmark position as Committee Chair. Through your chairmanship, Dr. Howell, the babies in our country today fare far better than they did before you arrived. A sea change has occurred.

You set sail with your motivated crew through uncharted waters, determining an effective

path forward. It was not long after you stood at the helm that this committee had a uniform panel for newborn screening and a plan as to how the panel should be expanded. Prior to this accomplishment, it was each baby for itself in the states, some faring better than others. Through your vision and unmatched efforts, we have sailed to smoother waters, erasing many of the discrepancies in the states, thereby minimizing the negative effects on our American families.

For more than seven years, I have attended these meetings along with my colleague, Nicky Gartsky. We have listened, questioned, studied and have been inspired by you on so many levels. Your patience to be available to answer questions means only one thing to us: the well-being and improved health of American families are at the top of your mind.

To explain how much we appreciate the support you have given us when answering all of our questions can be summed up in one word: priceless. Your patience and availability has also enhanced our principles and knowledge to do our part to create the

very best possible avenue for advocating greater awareness of newborn screening so that more education is possible to all American families.

Your words and wisdom will continue to inspire us as we move forward in this new era of newborn screening. You will be sorely missed here, but we know you will continue your good work in many ways. And we look forward to continue working with you on our efforts. Thank you.

DR. HOWELL: Thank you very much, Jill.

(Applause.)

DR. HOWELL: And I think many people will find the video that's been prepared by Jill's group to be a very effective educational tool, et cetera.

Next, we'll hear from Anna Marie Saarinen, who is representing 1in100 Newborn Screening. And Anna Marie arrives today -- do you want to come up and sit down -- after a very exciting letter concerning one of her passions, arrived yesterday.

Anna Marie?

MS. SAARINEN: Thank you, Chairman Howell, Committee. My comments that I had planned for today

changed yesterday at 4:00.

(Laughter.)

(Applause.)

MS. SAARINEN: Thank you for all your eloquent introductions, by the way. We're so jealous of your vocabulary, Dr. Howell. You should have your own Rosetta Stone (inaudible).

In the past few months, those of us who've been, sort of, working on this critical congenital heart disease issue have met with nearly 80 congressional offices to share information that has been learned and developed and provided via this committee and the evidence review process and the work group process. An additional dozen or so informational briefings were provided to HHS, HRSA, and other stakeholders that, I do think, moved the needle on an issue that had a lot of divisiveness. Information overcomes a lot of things.

We've also worked with the New Jersey Department of Health and the Implementation Work Group and established pilot projects that, not only get more hospitals adopting newborn screening for heart

disease, but are encouraging the meaningful use of electronic health information exchange. So hopefully, we're accomplishing multiple things through this wonderful screening.

In the year since this committee voted to recommend newborns be screened for heart disease, more than a hundred additional hospitals have implemented the screening around the country. Pennsylvania has introduced legislation since we last met in, whenever that was, May. New Jersey's governor signed their bill into law, literally, the days after we met, or within a few days, at any rate.

Starting on August 31st, that state started screening every newborn for critical congenital heart disease. And that all happened in eight weeks' time, by the way. The reporting piece and the infrastructure piece was still being worked on.

But to give a state credit for being able to put together a program, look at the evidence that's been provided and the guidance that was provided out of many key people in this room, and how a state can translate that into an operational program that's

screening babies has been inspirational. And the Commissioner and Assistant Commissioner have been wonderfully supportive in that state. I hope it's a model for others.

In Minnesota, we're now screening a population of what will be 15,000 babies in the coming year. We've translated our educational materials into three different languages. And we're working with I.T. at the Minnesota Department of Health to support electronic results reporting.

In fact, we're meeting just now in the next couple of weeks. We hope to have the system up and running very soon that'll make it even easier for hospitals, not just to screen, but to be tracking their results, which is going to be really important, I think, for this committee to know about.

I hope this effort has reinforced something very important: that the work here reaches beyond metabolic screening. Today 11,000 babies are going to be born in this country. And 110 of them will be diagnosed with some sort of a heart problem. Eleven of them will die before their first birthday.

I know. I know, not just in my heart, but on paper that what you've done here is going to change that number. More babies will survive because of the work that you did and the leadership that's now been provided at the federal level.

My dad was diagnosed with stage four cancer two weeks ago. No daughter wants to hear from the doctors at Mayo Clinic or anywhere that we would have had more options had we known sooner. No parent wants to hear that, either. Please know that the work being done here helps so parents don't have to hear that as often.

On behalf of my family, 1in100, and the CHD community, the Newborn Coalition, I thank you all for your important work.

Chairman Howell, the work you've done will be recognized by generations. You leave some very, very big shoes to fill, Kobe Bryant-sized shoes to fill.

(Laughter.)

MS. SAARINEN: I hope those that come after you can follow you in your wonderful footsteps. I'm

not sure if the person who did that military discussion earlier -- I learned a lot from that -- is still here.

Oh, hi, Mary. I'm not sure if you knew, but a third of the military hospitals in this country are already screening with pulse oximetry. So kudos to the military hospitals for their leadership.

Thank you all. It's been a pleasure.

DR. HOWELL: Thank you very much, Anna Marie.

(Applause.)

DR. HOWELL: We're next going to hear from Dean Suhr, who recommends the street vendors for quick lunches; right?

MR. SUHR: Absolutely. The hotel food gets a little old after a while.

Well, good afternoon, committee and Chairman Howell. I'm Dean Suhr. I wear three hats today, that of the parent of two children with a rare disease, one of whom passed away about 15 years ago, the other who I gave up her birthday to be here with you tonight -- this afternoon. But she is still with us. And that's

metachromatic leukodystrophy.

My wife and I formed the MLD Foundation 10 years ago. And we focus in on that rare particular disease. But today I want to start my comments in a new role that I have as the COO for the R.A.R.E. Project, a global genes initiative. And I want to acknowledge the work that this panel has done and Chairman has done for rare diseases since its existence.

Twenty-five meetings, seven or eight years -- I didn't come to the first meeting, so I don't know when that was. But you've come a long, long ways in that timeframe. And it's been something that I've observed and now have some responsibility to be more engaged in. And I just really want to acknowledge that.

The committee, under your leadership, but, certainly, with a lot of individual and group contributions outside of the scope of the people we see around this table, just really needs to be acknowledged. You've established the process. You've established standards. We heard about evidence-based

review. You have a methodology for making decisions, going forward.

Certainly, it's not perfect. Certainly, you'll get pressure all different directions as we look at the evidence. But you do have a process and a procedure.

And I think the results of that are validated by the 50 states and where we've come over these last seven years. The fact that those states, who have their own ability to make decisions, have honored what you've said and respected what you said and learned, based on that, I think, is a validation.

Clearly, parents are all for screening. There's no question about that. But when we get a little less emotional about that, I think the states really say it for us.

Specifically, for Dr. Howell, I've had occasion to meet him and talk with him and actually videotape him at a number of other venues other than this. And he's just a wonderful.

You're accessible. You're open. You communicate well. Somebody already alluded to your

sense of humor. You have a way of dealing with very complex issues in a very, very concise and friendly way. And that's really important, literally, to the millions of families out there that are the beneficiaries or are anxious about what this committee decides. And I just want to acknowledge that.

On behalf of the MLD Foundation and metachromatic leukodystrophy, we're not on the docket. We're not at the point where we have a diagnostic screen. There's much debate about the effectiveness of therapies. But we have a lot of challenges in front of us.

But again, we're going to be the beneficiaries, I hope, at some time in the relative near future of the process and the procedure you've put together. When we can show the evidence, when we can deal with and wrestle with the issues and the waiting that you have built into an evidence-based system that includes, in essence, variations at the ethics, the tradeoffs that aren't quite all numbers-based and the waiting, we're going to be the beneficiaries of that, as are many, many other

diseases.

And I just want to thank you for all your work, those of you that are going off. I challenge those that are stepping onto the committee.

And, Dr. Howell, particularly, thank you for your leadership.

DR. HOWELL: Dean, thank you very much for those kind words.

(Applause.)

DR. HOWELL: I'm told that Celia Kaye is back from lunch. It must have been quite a lunch.

(Laughter.)

DR. HOWELL: But if -- and Celia, of course, is the Czarina of the Mountain States Regional Genetics Collaborative Center.

(Laughter.)

DR. HOWELL: And she's going to have a few words to say.

MS. KAYE: I have a very few words to say. I was thinking I would get to say them from back there.

DR. HOWELL: Actually, the other thing that

some in the group may not know is that Celia was Chair of Pediatrics in San Antonio when I was Chair in Houston. So we have many bonds.

MS. KAYE: I know.

DR. HOWELL: The Texas bonds.

MS. KAYE: The great state of Texas, absolutely. Well, I want to thank you, Dr. Howell and committee, for this opportunity to say a few words to thank you all for the service that you've been rendering.

As Rod said, I'm Celia Kaye. I'm Project Director for the Mountain States Genetics Regional Collaborative Center. And on behalf of the Mountain states, particularly, I'd like to thank all of you, and especially Rod, for the leadership that you've shown.

I think we all are extremely conscious of the impact that the approval by this group of the uniform panel and the expansion of the uniform panel that happened through this group has made a tremendous difference in the way that newborn screening is thought of and taught throughout our various venues.

As an on-the-group person, a Mountain states person, I want to emphasize that in my few remarks.

What this group does really matters to the states, to the public health departments, to the community clinics, and, as a medical school person, to our medical students, our nurses, our physician assistants. They actually know what this group is doing. And I think the good example is the going viral of the ACCCHD recommendation.

I have had multiple e-mails about that since it happened, what, 24 hours ago, because people are interested in what's happening. They know that it makes a difference and that it will impact lives. So, again, from the regional collaborative perspective, from the on-the-group perspective, where people work every day and where differences are made in lives every day, I want to thank you for what you've done.

Rod, in particular, we so much appreciate your calmness, your humor, your focus, and all that you've done for all of us in the Mountain states. We appreciate your visits. It was wonderful to have you come and spend time with us, interact with

geneticists, family members, pediatricians, laboratorians. That matters.

Again, it makes change happen when people take their time and use their influence to actually see that change happens on the ground level. So thank you to all of you and looking forward to all the good things that are coming.

DR. HOWELL: Thank you, Celia. You're doing a great job out in the Mountain states.

(Applause.)

DR. HOWELL: We have Lori Williamson Dean next on our agenda. Here comes Lori.

MS. WILLIAMSON DEAN: So, Chairman Howell and distinguished committee members, my name is Lori Williamson Dean. I'm the Program Manager of the Heartland Region. And both Dr. Klaas Wierenga and Brad Schaefer send their regards to you.

The Heartland Genetics and Newborn Screening Collaborative thanks you, Chairman Howell, for your leadership and dedication to the work of this committee since its inception. The eight Heartland states have screened for the core panel of conditions

since July of 2008. And states are adding the LSDs and SCID disorders in the coming months.

Without the hard work of those who envisioned the regional collaboratives as a way to reduce disparities in access to quality genetics in newborn screening services across this nation and without your leadership to implement that vision, I know that the great states of North Dakota, South Dakota, Nebraska, Kansas, Oklahoma, Arkansas, Missouri, and Iowa would not be where they are today in terms of access to high-quality newborn screening and genetic services.

You've made a real difference in the lives of families across this country and in public health genetics. Thank you, Dr. Howell.

DR. HOWELL: Thank you very much, Lori.

(Applause.)

DR. HOWELL: And Jennifer Miller is next on our agenda. And Jennifer is the mother of Logan Miller.

MS. MILLER: Hello, and thank you for giving me the opportunity to talk to you today. I would like

to introduce a new disease to your list of heritable diseases. And it's called adrenoleukodystrophy, otherwise known as ALD.

Logan Miller is nine years of age. And we need the standard procedure for health care for children to 10 years of age to change. It should remain the standard procedure for small-town PCPs to ask for genetic screening called blood spotting.

We live in Pennsylvania, in Bellwood, Pennsylvania, very small community. And this is a very rare disease. One in 20,000 children, actually, have it. But 1 in 100,000, actually, are being diagnosed correctly with it.

So adrenoleukodystrophy is the disease. The abbreviation is ALD. We'd like to have this happen, and it's wonderful to hear that your committee is already tackling blood spotting and all the wonderful things that I've heard today that you do.

Logan's story began on 8/23/2010. He was struck by a truck in Bellwood, Pennsylvania. He was (inaudible). Due to the multiple facial fractures, he was put into Children's Hospital in Pittsburgh,

Pennsylvania, where an MRI was (inaudible for a few words.) They discovered, in addition to this life-changing event, that he was diagnosed with adrenoleukodystrophy. And that was on 9/22/2010.

This is an X-linked chromosome disorder. It's hereditary. And he had been born with this. So you can imagine how devastating that was for us, within a month's time, to realize this disease and not really understand it, but then, also to be -- where do we go from here? And what are his life expectancies?

So until this point, we knew nothing. We just thought that he had ADHD. And Logan had been asymptomatic, of course. So he just had the minor behavioral disorders when we were in school. So imagine how these educators feel when they have to deal with a child that has something else as devastating as this disease. And I'd like to tell you a little bit about the disease and what it actually does, that we've learned in a short amount of time.

But it meant when it's asymptomatic that it's presenting on an MRI. Adrenoleukodystrophy is a disease that is hereditary, of course, and a genetic

X-linked chromosome disorder. It's passed down. My biological father had it, however, I never really knew my biological father.

And this is a common story, that I understand, from -- we went to the Mayo Clinic in Minnesota in our travels in a short amount of time to try to get a transplant. And then, it had progressed too far, this disease. So then, we went to Kennedy - Krieger Institute in Baltimore, Maryland. But in order to spot this, we need to have the blood spotting genetic testing starting at 0 to 10 years of age.

A couple of (inaudible) after that, in order to watch the progression of the disease, we need to couple that with an MRI and very long chain of (inaudible) blood tests to be actually found as well. These children are being diagnosed with ADHD, bi-polar, Addison's, multiple sclerosis, which is, in fact, what my father had. All his life he thought he had it, but he really had AMN, which is actually the muscular version of adrenoleukodystrophy.

So his brother also had it. In the time that we learned, in this short period of time, the

school district wanted to get me involved with a support group. And I actually said yes to that, wanting to learn a little bit more about their experiences.

In fact, through that phone call -- we made one phone call -- that person was actually -- the numbers didn't add up in Bellwood. Bellwood's such a small town, so how could there be two children in that town with the same disease. And, in fact, the only way that can happen is if you're related.

Turns out that that person was my first cousin. And that child died in 2005. So it's very important, and it's a wonderful thing that your committee is actually offering to take this role and do this in all the states. So we appreciate that.

How can small town doctors, actually, in life situations -- my insurance would not allow us to have an MRI for Logan unless there was a traumatic reason to have it. So, in our area, the child that was before Logan actually didn't even have it diagnosed until after he passed on. And he had been diagnosed with all the things that I had mentioned

prior to this.

So we are actually here, one, to introduce the ALD Foundation. And we put it in Logan Miller's name. I actually have a picture that I gave them that was from the Caring Bridge Web site from Minnesota that I didn't see that it came up. And that's okay. But I also have literature from Dr. Westin Miller. And he works for the Mayo Clinic in Minnesota. I also have literature on the disease from Dr. Gerald Freeman. He worked under the Mosurs at Kennedy-Krieger Institute and John Hopkins in Baltimore, Maryland. So I'd like to enter that literature for you as well.

I would have had it already in your Web site, however, my e-mail address -- it doesn't recognize -- I have Hotmail, and it recognizes Yahoo and different ones. So I apologize for that. But I wanted to make sure that you get that information as well.

So, at any rate, we were given -- in our travels, we went to Minnesota in hopes of stem cell transplant. And then, last year, in October to

November, it actually progressed too far. And so, he was ineligible for that procedure.

So they gave him 18 months to 2 years. And that was 9/22 of last year. So thank you for your time today. And thanks for all your good work.

DR. HOWELL: Thank you very much, Ms. Miller. At the very earliest committee meetings, one of the presentations that we had was from the late Dr. Hugo Mosur, who was a leading researcher in ALD. And he discussed, at that time, the state of affairs with adrenoleukodystrophy. There have been a lot of progress since then, both in the diagnosis and therapy. So one would hope that this condition might be renominated at this point in time. It was never formally nominated. But there, certainly, has been a great deal of progress in that area. And it would be worth, certainly, thinking about that at the future. So thank you very much for coming and telling about your son.

MS. MILLER: (Inaudible.)

DR. HOWELL: Thank you very much.

MS. MILLER: Thank you. It was a pleasure

working with all of you today.

(Applause.)

DR. HOWELL: I wonder if Jennifer Garcia has returned from lunch. Thank you very much.

Is Natasha going to make the presentation, also? Or is that that you have covered all of -- you are a team today?

And then, I have Jim Bialick from 1in100 Newborn Screening.

Jim?

MR. BIALICK: I know that we're short on time, so I'll go quick. My name's Jim Bialick. I'm Executive Director of the Newborn Coalition. And we were, obviously, thrilled with the Secretary's letter yesterday and how much it was picked up. I know Politico ran with it. So that's always really good to see.

The one thing I want to talk about is just, kind of, how, with this recommendation, how we're starting to see some convergence of worlds here, where you're seeing something like a point of care examination, which has a lot of resonance in the

process within HHS for a lot of electronic health records development, even to the point where public health reporting can be -- can qualify for the lab reporting requirements of certain hospitals and providers.

The one thing that I want to point out, though, is that, in this ecosystem that we're developing here, there are a lot of blind spots. And one of those that I am seeing very frequently has to do with public health. And recently, there was a big HHS press event around Blue Button, which was this ability to spur insurers and hospitals to provide an entire patient's record all at once. It was, kind of, this big (inaudible).

And there was another announcement they made, which, kind of, got overshadowed, but I think has a lot of relevance here, which is that HHS is announcing, you know, another acronym, Advanced Notice of Proposed Rulemaking. So we're thinking about doing something about thinking about doing something.

And what you have there is that it would require that all individuals have direct access to

their lab results. And I know that this is going to have an interesting impact on newborn screening. And I know this is going to have an interesting impact on a lot of state laws.

And so, you know, I definitely suggest that, maybe through your associated organizations or through this body, that comment be made on that, because I think that, where the thinking is is that this is information that's going to come from the labs directly. And so, especially with newborn screening, especially with something that is -- you know, has had a lot of debate about that, you know, a lot of standards about that, it's going to become increasingly important that that information -- you know, that there be a consensus on how that information is managed.

So I just, kind of, wanted to put that on the radar as well as talk about, you know, how these things are starting to converge a little bit. And it's really an interesting, exciting time. But I think that it's going to take the input of a lot of knowing people that we have around this table.

DR. HOWELL: Thank you very much, Jim. And we'll look forward to --

(Applause.)

DR. HOWELL: That completes all the persons that I have on the -- who has signed up for public comment. And surprisingly, we're back on time, which is remarkable, but since we had gotten so far behind. And so, we'll now move into the next phase or activity. And Sara is going to talk about the agenda and the plan for the subcommittee sessions that will follow our break.

DR. COPELAND: Thank you. This will be very, very fast, not 15 minutes, by any stretch.

You will notice, as you go to the subcommittees today, that they will have very similar agendas. And the idea being that we would really like to use this time of transition to, kind of, first off, enumerate what you have already done, take an inventory of what is ongoing, because we will have many subcommittee members who are going off and new ones coming on. So it would be nice to know where we stand and possible future roles of the subcommittee.

And one of which, and not a small one, is whether or not you should be a standing subcommittee or maybe an ad hoc subcommittee and whether or not we need to consider other subcommittees, much like Ned mentioned earlier. So this is a time of reflection, but also planning for the next stage in the Advisory Committee and just to remind you where you will be.

The Laboratory Standards and Procedures will be in City Center 1. The Follow-up and Treatment Subcommittee will be in this room. The Education and Training Subcommittee will be in City Center 2.

MALE SPEAKER: (Off-mike.)

DR. COPELAND: I have no idea.

MALE SPEAKER: (Off-mike.)

DR. HOWELL: (Off-mike) I think so.

DR. COPELAND: Yeah, then --

MALE SPEAKER: (Off-mike.)

DR. HOWELL: Yes, I think (inaudible).

DR. COPELAND: Okay. So that is it for the agenda.

DR. HOWELL: Okay. So the schedule calls for us to have a break at this time. And the

subcommittee meetings convene at three and end promptly at five, as you can see. And then, tomorrow morning, we will start again with the continental breakfast of the committee at 7:30 and hear from the subcommittee reports beginning at 8:30. So off we go.

(Whereupon, at 2:35 p.m., this session of the Advisory Committee adjourned.)