AFTERNOON SESSION

CHAIRMAN HOWELL: Ladies and gentlemen, we need to get moving along here. We've had so many exciting things going on today that we're quite behind time.

We will end promptly at 3 o'clock, as we always do, but today there's even a greater sense of urgency because, obviously, Yom Kippur, is rapidly approaching. And certainly the folks who have come locally need to get out of here. Unfortunately, some of the people nationally were not able to join us.

Dr. Kaufman, who has laboriously driven down from Baltimore, has agreed to have his presentation on the efficacy of newborn screening from a family perspective -- we will hear from him later. He's agreed to not present this afternoon so that we can stay in a timely fashion. But we will hear from him later.

But we will ask Sue Berry to start with her presentation about the access to medical foods

and formula, a survey of regional activities. Sue, could you come? Sue, as you know, is Professor of Pediatrics and Genetic Cell Biology and Development out in Minnesota and the source of most of our public comments on various and sundry things. She's going to talk to us about this effort that she's been working on. Sue? She's going to present data I think from region 4, region 3, and region 2 about her survey.

DR. BERRY: Well, to be more correct, it's not really just my survey. It was the survey of a really excellent workgroup of the Follow-Up and Treatment Subcommittee, and I want to acknowledge the strong support and advocacy of that group in providing this forum for our being able to learn a little bit more about medical foods. And so I'm speaking on behalf of the Medical Foods Expert Panel, which was a sub of the subcommittee. It gets kind of intricate at times here.

I also want to acknowledge at this point the expert participation and the hard work and

analysis of Mary Kay Kenney from HRSA who did a lot of the data collection encouragement and analysis and worked very hard on this. Much of this is due to her hard work as well.

So all of you are familiar with the committees, but each one of them has a charge. The main charge that we were engaging in looking at in this was to work on our charge to develop recommendations for overcoming identified barriers in order to improve short- and long-term follow-up of newborn screening results. In an ironic sense, you don't get much improvement in newborn screening results if you don't treat them correctly, and I think that that's one way of thinking of this. This is also an accountability issue for us.

We hear frequently from families that share their concerns with us. They are very moving and important, and for me personally as a practitioner, I think this tells us a lot about why we do what we do. This was a comment from one of the surveys that we got, and I'm just going to read

it to you so you can hear this young woman's voice.

She said: "I do not think it is fair for children with medical problems and you do not get help to provide for your child. I have a child with LCHAD and I am very short for money and unable to work due to my child's needs. I am getting help on medical bills but not for her MCT oil that she needs for the rest of her life or she may not survive. I currently have that medical in collection because I am unable to pay for. What am I supposed to do to pay for it and keep getting her medicines? Also it is very expensive to buy the foods she requires. I do not get help for her fat-free/low-fat foods. Please help pass this on through States and put yourself in our position. What are you to do?"

While we will not be able to correct all of the problems that she explicates here, I think what she tells us is that the need is very real, and this is a voice that I thought was important to share with you.

So what really is the problem that we have? To frame the issue, medical foods, which are often referred to by families as special formulas or protein substitutes really aren't drugs, but they are substances of nutritional value.

Medical foods aren't optional. They are the treatment for the inborn errors of metabolism that we have worked to screen.

And treatment is lifelong. It doesn't just happen and then it goes away and you fix it. People have these diseases all of their lives.

Everyone needs foods, but traditional foods can be harmful to persons with inborn errors of metabolism.

And medical foods are substantively more expensive than traditional foods.

Because they are foods, they are excluded from coverage by many insurers, and so the costs to the family may be prohibitive. Coverage is at best variable.

Affected persons cannot survive without

medical foods, but they can't afford to buy them. That's a terrible horn of a dilemma to put our families in.

We're going to talk more about the definition I think hopefully in the discussion regarding this, but there is a formal definition for medical foods: "A food which is formulated to be consumed or administered enterally under the supervision of a physician and which is intended for the specific dietary management of a disease or condition for which distinctive nutritional requirements, based on recognized scientific principles, are established by medical evaluation." It sounds concrete, but there are some real gaps in how this can be applied that have limited the ability to get medical foods paid for. This was specified in the Orphan Drug Act, and I'm pretty sure we're not going to change the Orphan Drug Act, but we may need to have another strategy to get at that. So is the definition part of the problem? I think that will be one of the questions we have to

encounter.

So what are the nutritional treatments? Those are the subject of our survey.

First, medical foods. These are the formulas that I've just talked about. They usually supply a substantial portion of the nutrition for the treatment of the specific inborn error of metabolism. Typically they have a restricted amino acid component. That's the most common formulation for medical foods.

There are also supplements or sometimes referred to as nutriceuticals which are pharmacologic doses of vitamins or cofactors. A good example of that is the treatment of biotinidase deficiency is administration of pharmacologic doses of biotin. Some of the disorders result in specific amino acid deficiency, and patients need amino acids provided as substrate to complement their metabolic condition. There are also other vitamin-like drugs such as carnitine and things like MCT oil that are essential treatments

for certain types of fatty acid oxidation disorders.

There are specially manufactured modified low-protein foods. These are not your average mother's low-protein foods. These are really restricted in the protein content and very specially prepared and provide the ability for a child to have a much more normal lifestyle.

Now, medical foods require physician supervision. They're all essential elements of therapies for treatments of inborn errors of metabolism, and many families, in addition, require medical equipment and supplies that are needed for feeding. Those also fit into a problem for families that we wanted to address in thinking about this issue.

So to do this, we took a two-part approach, and I think we've previously discussed what we found in our Medical Foods Workgroup meeting. In June 2008, we had a meeting where we got together with representatives of insurance

companies, along with the expert group, and got some information and resources with regard to our understanding of the problem at hand.

The problem in part is this. Each insurer has their own practices. Private insurers, public insurers using private vendors, self-insured plans, employer-based plans -- each of those has a different set of rules, and each insurer has their own practices responding to those rules. And public practices also vary from State to State. Moreover, each policy, even when you have the same company, can have different coverage. Contracts can result in different coverages for the same insurer. It's kind of mysterious sometimes how they make those decisions.

Each State has different rules or laws covering provision of medical foods which also impact the availability. Those were beautifully summarized previously and presented to this group. So you can go to that link, if you want to. But even when laws exist, they may not cover all

insurance carriers. For example, ERISA and self-paid insurance is a good example of where there are a lot of exceptions for these. And even when laws and guidelines exist, they're subject to interpretation. So we have a lot of problems with the payment side.

To try and get some better information about this, this group undertook a medical foods survey. We decided to do a parent survey for insurance coverage of medical foods for children with metabolic conditions, and that’s what we're going to talk about today. This is a project that took about two years to get done. We just thought we’ll just send out a survey. No problem. It doesn’t quite work that way, but that’s okay.

So what were the objectives of our medical foods survey? Well, first we wanted to survey parents of children -- and we’ll come back to that as a point as well -- with metabolic conditions to look at their current coverage and the actual coverage for the medical foods and the

materials needed to administer them.

Our rationale was to inform federal and State public policy decisions and this group to try and reduce financial barriers for families that were needing treatment.

What kinds of information did we seek in this? Well, we wanted to know what the needs of children were for medical foods and formulas, modified low-protein foods, those prescribed supplements, and the supplies that were needed to administer them.

We wanted to know how much families spent for these things, what were their out-of-pocket expenses, and what proportion of expenses were paid for. We realized this to a varying degree, but I think we got some very important information.

We established our expert panel, and we undertook initial cognitive interviews to make sure we were asking the right kinds of questions and that families understood what we were asking. And those took place in two cognitive interviews.

We did some pretesting survey validity and reliability in the fall of 2008 at three sites where we gave people the instrument we were going to use and then modified it.

The survey asked about the child's diagnosis. They asked about the health plans covering the child care because a lot of people have more than one way to get foods and other things covered. They asked about what materials were used by the child. They asked the extent to which those items were covered by their health plan, including dollar amount per month. We asked for an estimate of monthly out-of-pocket expenses, if not fully covered, and if health plans had caps on coverage.

So what we had to do to do this is we implemented this with -- this is a survey of convenience. That's one of the caveats that I'm going to share with you. We were able to obtain data from people who volunteered to help out. And we undertook this with a collaboration of the NYMAC

region, region 2; the southeast region, region 3; and two centers in region 4, with support from the National Newborn Screening and Genetics Resource Center. And we undertook the survey in the spring of 2009.

We targeted this at children. We didn't target adults, and that's an equally important group but a smaller proportion. So it was harder to get solid data, and we did not undertake a survey of adults.

We received responses from 305 families across the three regions.

We did this following an IRB process. We received approval for implementation in various ways. Typically these were expedited reviews. We did the paper survey that was administered in the genetic centers, and the responders completed the survey anonymously, which helped for our expedited review. We didn't collect, for that reason, tons of demographics, but it makes it a little harder to go back and double check all the data because it's

anonymous. But we knew the State of residence, the age of the child, and the diagnosis of the child.

Each of the genetic centers submitted data to their regional collaborative for central evaluation and coding, and then the regional collaboratives, in turn, had that data to inform their own planning and submitted the data on up to HRSA and to Mary Kay for further analysis and integration.

I want to acknowledge the centers that participated in this activity under the leadership of Cindy Cameron in region 4, Kathy Harris in NYMAC, and Ronnie Singh in the southeast region. We really appreciate the clinicians who participated in this activity.

So the data. Here the blue is the youngest children, the children between 0 and 5, and this is a summary of the distribution of the surveys done in each of the three regions. In some cases, I'm going to show you data that will sort of look by region, but this is the only place where we

split it out so you can kind of see where the information came from. About half the children in this group were children under age 5.

When we looked at health care coverage, about 25 percent of the children actually had more than one funding source, and only 3 children really didn't have insurance, which is actually better than the national average, if you think about exposure of children to insurance failure. I think that's probably because once they were identified as having such a critical disorder, in many cases other strategies were found to get them covered. So that was a point of cautious optimism we had for this.

A particular area of interest for all of us was the utilization of WIC, and it turned out that 30 percent of families with children under age 3 used WIC. So that was very important. I didn't break it out here, but I wanted you to know that WIC was very important.

These are the total number of children

using each type of coverage. So if you added them all up, it's going to be more than 305. Many children had more than one funding source.

The next slide is a busy, difficult -- I debated about even putting it this way, but I'm going to try and go through it so you know.

In the upper left-hand corner, the first pie you see there is for medical foods, which was the main target of our study. The big blue slice that you see is the people that obtained their products from a pharmacy, and to a large extent, most people, if they get them from a pharmacy, they're typically paid for.

The next big red slice next to it is from county and State health departments, and in many of the States, foods are supplied through the county or State health department. In those cases, most of them are paid.

The next two slices, however, the green and purple slices, are the manufacturer or the Internet. In those situations, most of the time

people have to pay for those out of pocket. So you start to begin to see some of the problems.

The next two slices are medical supply and home health companies. Many times those are also paid for, but when you go on around to the hospital or clinic, it's paid to varying degrees, and then WIC I think is a good source for many of the families.

When you looked at modified low-protein foods -- and the reason I mentioned the Internet is something where it may not be paid for -- here that great big green slice for the modified low-protein foods is from the Internet or the purple one next to it directly from the manufacturer. That tells you a little bit about the scope of the problem just from that one thing alone. Many families have to go direct to these sources, and they pay out of pocket to pay for those modified low-protein foods.

For dietary supplements, most of those ended up, by a large majority, being supplied by pharmacies. There are still problems with getting

those paid for, but in many cases if they come from pharmacies and are prescribed, they're paid for. So that’s a point of impact, but not as big as it is for either medical foods or modified low-protein foods.

And in my own sort of half-thinking, though I know it’s not really true, I thought of feeding supplies as something that surely must be paid for. As far as we can tell, the sources that are mostly being used to gather feeding supplies are things that would be typically paid for by insurance, and that was what we ended up finding out when we looked at the actual numbers of what got paid for.

So impact here. There are two places where a major impact in terms of the kinds of places where they won’t get paid for and that’s medical foods and modified low-protein foods.

Now, most of the children ended up needing to use lots of products. 50 percent of the children in this group actually used feeding

supplies. So the way that this is set up is that these are the number using the product, and of the children in the group that we selected, 150 of the 305 used feeding products. So that's what adds up on that bar for that one, for example. 71 percent of them used dietary supplements of some sort. 60 percent of this particular group used modified low-protein foods, and 84 percent of them, since it was a medical foods survey, used medical foods. We took comers who weren't always using medical foods but used the other things so that we could have -- that was just how they got handed out. That's who filled out the survey. They answered as they answered.

Now, almost all the children, 80 percent, used at least two of the surveyed products. That's what this pie really shows you, that many of the children used these products on a daily basis and they used more than one of these. So they have a comprehensive and wide need for these products in their full forms.

This is, I think, the graph that I found to be most important and I thought was kind of the most upsetting to me in some ways. So the way that I've got this set out so that you can see it is the total number -- we asked them to tell us all the ways that they got things paid for. So the answers, since they may have used three or four sources, add up to way more than 305. It's all the various sources that they used.

And for medical foods, of the 296 responses that engaged about medical foods, because they were using multiple sources to try and cover this, we were really -- it's good to see that private coverage and Medicaid and State coverage covered many of these. But there's still a significant fraction of people who have expenses out of pocket for paying for medical foods, and that was true for supplements and most strikingly for modified low-protein foods. 172 families that provided a response and talked about how they paid for modified low-protein foods ended up having to

pay some or all of it out of pocket. So we found that to be quite striking particularly for modified low-protein foods.

In terms of the actual costs to the family, families really had a hard time giving us an actual dollar amount they spent. I'm not sure if we didn't ask it right or it was something that was hard to know. But we do have a surrogate for that. We asked them to tell us a range of what they've paid per month for the various attributes of these. And here, the good bars, the happy bars are the orange and blue ones. If they paid nothing or between $0 and $100, those are the orange and blue bars. Anything below that, however, except for the "don't know" bars -- who knows what those are -- represent funds that families paid per month for the various products. Here we lumped supplies and the supplements together because those were more likely to have been paid for and really focused on the modified low-protein foods and the medical foods as points of risk for families.

And you can see although many families paid little or nothing, some families paid quite substantially. The sort of number -- I did a calculation where I used the mid-range of these values and then calculated how much as a total estimate divided on everybody in the group, if you looked at how much they would pay, the average family would pay $3,800 per year. And since you can see that many families paid little or nothing, some families are paying a lot.

So what did we end up learning in the end? Well, we found out to our pleasure that nearly all the children in this group had some type of health care coverage. That was a good thing. Unfortunately, it didn't always pay for these products.

Most of the children we surveyed needed more than one category of food or supplies, and that makes sense. You don't just feed kids formula. You feed them real foods, and you make sure that they have the supplements they need to

complement those. And if you need to use a feeding tube, you use it. So most of the children needed more than one category.

Coverage was variable, but there was at least some out-of-pocket expense for about 20 percent of the families using medical foods, for about 30 percent of the families using supplements, for about 35 percent of the families using feeding supplies, and for about 60 percent of the families using modified low-protein foods, they had to pay out of pocket.

So what we do know and what we don't know. We didn't differentiate this by diagnosis. I think that because many laws specify PKU as an individual disorder, and we had a lot of PKU families in this. We may have overestimated the number of people who get things paid for. I think the problem is even worse for families who don't have PKU. That analysis can be done. I just didn't do it.

We didn't separate by age to any degree

except to make the notification that WIC was important for younger children.

We didn't have much luck finding out if families were capped on insurance, and that was part of the problem. They just didn't know the answer to that.

They had a hard time telling us their out-of-pocket cost as a specific number so that data was terribly incomplete. We worked hard to get it. Mary Kay particularly put a lot of energy into trying to help people do it, but it was very hard to get.

We found out that need-based supports were very significant resources. So WIC was a very important source of support for families and that Medicaid was critical. In fact, ironically it's a lot better if you're so poor that you have to be on Medicaid because then you can get your medical foods paid for. Medicaid plays pretty well, but otherwise it's a problem.

Modified low-protein foods are

particularly poorly supported. WIC doesn't pay for those.

And although patterns of coverage varied a little bit from region to region, all the regions observed significant challenges for families in paying for the products.

So what's happened? You guys are very familiar with this because this group has been very engaged in this. This committee has communicated already three times with the Secretary regarding medical foods, first in the letter of April 7th where we had an interim response. There may be some more updates than what I have on this slide. So I apologize if I haven't got every piece of this correct.

There was a subsequent letter in March of 2010 where we looked at gaps in insurance coverage and specifically mentioned medical foods as an important gap.

We had a letter in June where we updated our recommendations with regard to medical foods in

light of health care reform, and there's an interim response saying we're going to get a response in a timely fashion, which we always have. So I expect that as well.

Also, I think one of the outcomes of our interest in this and the hard work of families and advocates was to begin work on the Medical Foods Equity Act. I think many of you heard about this. It addresses a number of the issues that are relevant to this, but as you know, legislative measures can be quite a challenge to undertake, and this will be a process, not an event, to have changes in legislation.

So what next? What next for this group? What next for our subcommittee? Obviously, we're going to very much anticipate the results from the Secretary with regard to the questions we've already posed, and I think this is going to be a very important avenue for advancement of this.

We, of course, are going to monitor the progress of the Medical Foods Equity Act and the

benefits package that we've heard a little bit about in our discussion this morning, most notably the idea that medical foods may need to be made an essential benefit to be able to have the action that we need to take place in this.

I am delighted to have our colleagues from the FDA join us today, and perhaps they can give us some additional insight on the process by which in working with FDA to think a little more precisely about how we might define the needs and products that are necessary for treatment of inborn errors, if we might in regulations or rules find ways of some relief as well. I'm hoping for their comments, with permission of this group. Since they came here to tell us about it, I'm happy.

And then for our subcommittee, obviously, we talked about whether we needed more data, whether we wanted to include other regions, if we want to include other ages, if there were some other things that we needed to do to sharpen our argument. And also, I think we should publish this

information to make it more broadly known.

But we stand prepared as a group, I think, to work harder on this.

At that point, I'll open this for questions and you guys have a good discussion about this because I'm not an unbiased observer.

(Laughter.)

CHAIRMAN HOWELL: Thank you very much, Sue.

I wonder if Tim, Dr. Cote, from FDA would be willing to come up and discuss the issues of medical foods, among other things.

DR. COTE: Thank you so much, yes. My name is Tim Cote again. I'm a physician. I'm the Director of the Office of Orphan Products.

The term "medical foods" is defined and codified in law in only one place. That's in the Orphan Drug Act. That definition has proven to be near useless. It defines things so broadly that literally thousands of foods from prune juice for prostate health to wheat germ for prevention of

Alzheimer's meet the legal definition of medical foods. And nobody really wants to take on all the stakeholders behind those products.

So we have a problem here. The need that, Susan, you've outlined is very, very real.

I was relating earlier that this is a new field for me. I'm a pathologist by training. I have met some of these mothers, and to a mother, they complain bitterly that a product which is very much and very specific for the treatment of a very specific disease is not being paid for as drugs would be routinely.

The problem is that our agency is the group that needs to be able to identify that this is the product which is used for the treatment of that disease. And without such clear direction and definition, CMS is not really capable of deciding what to pay for. So we are going to be working with CMS on doing this. I myself will be feeding into the Secretary's response. I think you're absolutely correct that you will be receiving a

timely response back from the Secretary on this, and we are working closely on it.

To my mind, I think we need a subset definition that identifies that medical foods are -- excuse me -- that some new terms such as “metabolic product,” “nutritional products,” are a subset of medical foods which are specific for the treatment of these 29 diseases as identified by this committee. Moving forward, what we certainly will need to do is find a way to identify specifically what they all are.

So those are the kinds of directions that I will be giving back to my commissioner. My Deputy Commissioner, Dr. Josh Sharfstein, is a pediatrician himself. He’s apprised of the problem here. He has given me his support in moving forward. He understands that there's a definitional issue, and we will try our best to work with you closely to solve this problem, which really shouldn’t be here. We really should have solved this a long time ago, and it’s a terrible

burden for parents who have this issue and there's no reason for it.

CHAIRMAN HOWELL: Thank you very much. I guess one question I have is specifically how do we move forward to work with you about this new term, for want of a better word.

DR. COTE: Sure. Well, my experience with this administration is that it’s an extremely responsive administration. So you have a number of different avenues that you can look at.

First of all, you're going to get a response. You're going to get a rapid response. It’s coming. Okay? I’m actually going to be working on it today. So it is coming.

You have avenues of citizens petitions.

And the agency has some latitude in terms of its authority to promulgate regulations from existing statutes. The Orphan Drug Act is such a statute, and it has promulgated regulations related to that statute and could do so and define, for example, a subset of medical foods if there were a

reason to do that, and I think that there may well be. That process occurs when outside stakeholders request it of the agency, and the agency moves forward with its authority to promulgate a proposed notice of rulemaking through an established process in Federal Registers, through which comments are solicited and so on and so forth. Many of you here are very familiar with that.

So I can tell you that this is an area where the agency really wants to be responsive to a clear need, a bureaucratic problem that needs to be fixed and lives that hang in the balance. So I know that there are people at the very top of the agency -- me being a couple of levels down as a director of an office of 43 or so people, but Dr. Josh Sharfstein being the number 2 man for an agency that regulates a quarter of the U.S. economy. So I think we're going to get somewhere. I think the time has come.

CHAIRMAN HOWELL: Thank you very much. That’s very encouraging to hear that we're going to

make progress there.

Mike, you had a comment?

DR. COTE: And we will be at these meetings going forward. I'll make certain that somebody on my staff is here at every one of the meetings going forward.

CHAIRMAN HOWELL: Mike had a question.

DR. WATSON: I suffered from post-lunch drowsiness there momentarily. Is this just notice of conditions associated with newborn screening, or is this all the genetic diseases for which medical foods are prescribed?

DR. BERRY: I think there are a couple of things that we need to keep in mind. We crafted the letters, and I was very grateful to Michele for being as precise about this as she was and throughout for reminding of this. The way that it was crafted was to talk about the screened disorders and disorders as defined by this committee. I think that what the issue is, for example, is that you need the exact same products

to take care of kids with urea cycle disorders and just because we can't screen for them doesn't mean they don't fit under the same rubric. So I think we have to be very precise in making our request so that we don't leave out the big groups.

But the same point and one of the things we have to keep in mind is that we've got to get the biggest bang for the best benefit that we can, and if it means fixing part of this and fixing the rest later, we should do what we have to do to get at least a marginal change done. So if it has to be only about the 29, then it has to be only about the 29. I don't think that's the right choice, but let's think as carefully as we can about trying to include as much as we can that's appropriate.

That's my thought. I don't know if others --

CHAIRMAN HOWELL: The letter that was written to the Secretary specifically talked about the conditions for which screening has been recommended by this committee, but also clearly

pointed out other appropriate metabolic diseases. Now, the thing that we have not discussed -- and I don't know exactly how that would work -- is how do we decide that it's appropriate to provide nutritional material, corn starch for patients with type 1 glycogen storage disease or something of that nature. But that would have to be done if that would be the way this goes.

DR. BERRY: But I would think that the first priority should be medical foods and modified low-protein foods, and those other things would have to be worked on in my own personal view. I'm not the person who decides.

CHAIRMAN HOWELL: Yes, right.

DR. WATSON: That's good. Once you change the definition of it and move forward.

CHAIRMAN HOWELL: Sue has posed several questions for the committee as far as where they proceed in the future: number one, to extend the survey, to focus on elements of highest impact, and to publish. What's the wisdom of the group sitting

around the table on those issues?

(No response.)

CHAIRMAN HOWELL: It must have been a very good lunch judging from the amount of wisdom I see pouring forth.

(Laughter.)

CHAIRMAN HOWELL: Gerry?

DR. VOCKLEY: I think number 3 is important, so get the publication out and make this information more broadly visible. I think that's helpful.

I always hate to do more of the same, so I'm not sure that extending the survey is going to do much to move it forward. We've got a snapshot of the problem. Yes, it might vary a little bit if we go to other jurisdictions, but I don't think it's going to change us fundamentally. So I think moving forward, if we're focused on the elements of highest impact -- and I'm not sure how you're going to define those, but I think that's what you want to define. What's going to be worth your time?

CHAIRMAN HOWELL: Ned?

DR. CALONGE: Gerry, the only thing I would tweak about that is if there's a list of things that need to be approved, I think to the degree that we think about the entire list -- I'm just worried if you focus on the elements of highest impact, that's all that would be approved. So I guess I feel a little bit differently that we could have a prioritized list, but I think as much as we can be complete with our earliest recommendation, the better that would be.

CHAIRMAN HOWELL: Is there a sense that anything would be gained by extending the survey? In other words, you've done three substantive regions. I think you might have bigger numbers. Coleen?

DR. BOYLE: I guess I would like us to keep in mind the fact that we have the opportunity to use this survey as a tool to monitor the impact of any changes we make to the system. So whether or not we decide to extend the survey to other

areas, if there's concern in different regions to see that whether implementation of changes in policy, CMS practices, whatever has the same impact in others. It's at least a baseline from which to monitor. So if we did it, that's the context in which I would do it.

CHAIRMAN HOWELL: You don't think you could accomplish that by using the regions that have been surveyed and then look at --

DR. BOYLE: I would look to the regional folks to answer that question.

DR. BERRY: At least one of the regions commented to me that they felt that by extending it, they would have some additional impact in each of the States individually while we worked on this on a national basis. So some of them were interested in using it as a tool in that context as well State by State. If you've seen one State, you've seen another State in terms of how they work with these things.

CHAIRMAN HOWELL: Further comments?

We've heard that it would be wise probably to focus on all the elements, and you might prioritize those, et cetera.

If there's significant interest in publishing it, I think that should go ahead.

And then the survey. It seems to me that there's some sense that it would be advantageous to make it available and gather the other data.

DR. BERRY: We would be happy to make the survey available. We might make some changes based on our experience with it, but I think that the Coleen is right that it’s a good instrument for surveillance if we correctly use it.

CHAIRMAN HOWELL: Because if there is a substantial change in the reimbursement, it would be nice to see if you can demonstrate that. It might be one of the first controlled studies we've been involved with.

(Laughter.)

CHAIRMAN HOWELL: Any other comments for Sue? Fred?

DR. CHEN: It's useful to hear from FDA that this actually could get changed through the regulation process. So I assume our group is going to keep monitoring it. If it makes it through the Federal Register process, we would have opportunity for comment and we should certainly encourage that process.

CHAIRMAN HOWELL: Yes, and I think it's very helpful that we benefitted from having an FDA representative on the committee, but having this other office represented I think will be helpful. And we're very pleased that they're here today and have sworn to be back regularly in the future.

VOICE: I wanted to add and I want to emphasize this survey was done for ages 18 and under, and the problem is much more magnified -- as a clinician, I can tell you -- for older individuals even more. So if there's ever a need to document that data, that is something to keep in mind.

CHAIRMAN HOWELL: Have you thought about

looking at older people?

DR. BERRY: We actually considered that when we did the initial study, and then decided, particularly in our initial phases, that we would concentrate on the area of highest impact. But clearly, it’s magnified in adults. That’s 100 percent correct. Anything you're talking about happening in kids, multiply it times 10 in the adults. There's just not as many of them.

CHAIRMAN HOWELL: Jana?

MS. MONACO: I was just thinking along those same lines because many of the States, as we learned with PKU, do have coverage up to age 18, and then all of a sudden, those individuals drop off in their older coverage.

CHAIRMAN HOWELL: Well, maybe that information could be gained going forward.

DR. BERRY: Yes, we can certainly extend the study to encompass adults as well.

DR. CALONGE: I just am uncomfortable about the purview of --

CHAIRMAN HOWELL: We've been having a sidebar about the fact the name of our committee is infants and children, and most people are grown-up children.

DR. BERRY: Right. That's the only reason we focused on children in this. If you think about what's going on for kids, it's more in adults.

CHAIRMAN HOWELL: It could be accomplished, I think, through the regional collaborative network which does clearly have purview that goes well beyond the purview of this committee. I think this committee should do anything that's important, but that's a separate issue.

(Laughter.)

CHAIRMAN HOWELL: But the only thing that happens to you after you're 18 is you get older. Nothing else worthwhile happens.

(Laughter.)

CHAIRMAN HOWELL: Are there any other

things? You've done a lot of work and you've been working on this a long time. So hopefully this will be helpful and you can get this published. I do not think you should hold up your publication to start looking at other States, but you can acquire that going forward and focus on all the elements. And we're glad you're prepared to move further.

DR. BERRY: Always prepared.

CHAIRMAN HOWELL: Thank you very much.

DR. BERRY: Thank you.

CHAIRMAN HOWELL: I would like to take the committee discussion back to the -- we're going to go back to the discussion of the morning concerning the newborn screening using pulse oximetry for critical congenital heart disease. As you remember, Gerry had made a motion about enthusiasm for including this, and then there were other concerns about holes in the data and things of that nature. So Gerry and Jeff largely have come up with a recommendation for this committee to consider that would take into consideration many of

the things that you've heard.

DR. VOCKLEY: First, to remind you we had a request to have this grid back up. This is our category of recommendation grid, and based on the discussion prior to lunch, we're really in between categories 1 and 2. And if we go back to the SCID a couple of meetings ago, I think what we were really saying was that there's a sort of level 1 or .9 or something -- 1.1 maybe if we're going the other direction -- where everybody is really convinced it's a good idea, but we just need a little bit more to nail it down. That's as opposed to 2 which is it looks like a good idea but there are still some pieces missing. And this is why for this discussion, I made the motion to go to category 1 because I think it's there with a few pieces that would make us all feel more comfortable going forward.

So we put together just a statement that, again, really went back to the SCID recommendation that would be a modification of what was moved

before. And since I don't think I'm actually allowed to modify my own motion, maybe this is Jeff's modification. I don't know.

CHAIRMAN HOWELL: You can retract your motion.

DR. VOCKLEY: Well, whatever works. Let the minutes show I'm amenable to anything that gets us out of here.

(Laughter.)

DR. VOCKLEY: It looks like I may have cut off the top line of this which was that we recommend the addition of critical congenital heart disease to the uniform panel with the understanding that the following activities will also take place in a timely manner. NIH shall fund research activities to determine the health outcomes of affected newborns with CCHD as a result of prospective newborn screening. CDC shall fund surveillance activities to monitor disease incidence. Pass it around. HRSA is going to guide State health departments in the integration of

screening into their programs, and then HRSA shall also fund the development of, I guess in collaboration with public health, health care professional and family organizations, appropriate education and training materials for family and public health and health care professionals relative to screening and treatment of CCHD. That one was added quick on the fly.

CHAIRMAN HOWELL: So that's the nomination. Is there a second?

DR. VOCKLEY: This is now the recommendation.

CHAIRMAN HOWELL: And Jeff, you're seconding that since you helped write it.

DR. BOTKIN: Yes.

CHAIRMAN HOWELL: So we've had a nomination and a second. Now we'll have discussion of this recommendation. Ned?

DR. CALONGE: I have a number of comments.

The first is I don't know what we're

recommending. I don't know what cutoffs. I don't know what technology. I don't know what timing. I think it is difficult to add a recommendation when we don’t have the level of specificity of what testing we're talking about. I would hope that moving forward and included in the motion perhaps as a friendly amendment is we actually be specific about what it is we're recommending adding.

I saw huge variability depending on when you tested. I heard that there's variability in the technology that’s already changed. I heard about different probes, different sites. So one of the critical evidence items that was in the evidence report is how does screening test accuracy vary by the age of the neonate, placement of the probes, and threshold value for action. I don't see answers to any of those questions. So I’m uncertain I know what we're recommending.

We also have an evidence report that has as a critical evidence gap, what is the benefit of adding a pulse oximetry screen to infant outcomes

compared to usual care. That's a key evidence gap.

Now, I think whenever you do an evidence-based recommendation, you're evaluating the risk of being wrong, and I want to explain that in two ways.

One is what are the health risks of being wrong. If this is the wrong decision, what's the down side? From my standpoint, one of the best parts of the recommendation is that from an important health outcome standpoint, we haven't seen much evidence of down side. I don't think that the false positives are much of an issue if we time the screening right. I don't think we'll be over- diagnosing or over-treating. There could adverse events that we haven't thought about. Separation of the mom and baby at infancy is a bonding issue with significant impact that I don't want to overcall or under-call. But I'm just saying we can't say that there wouldn't be any harms associated with being wrong.

The other thing is what is our risk that

we'll be shown to be wrong later. So what's the real risk of this is the wrong decision. I would just point out that in our best intentions in American medicine, we have often been wrong, and I just want to make sure we understand that going forward.

Ultimately all evidence-based medicine decisions require a judgment, and the rules of evidence are the rules of evidence. But two groups of well meaning scientists have looked at the same evidence body and come to different conclusions because of the judgment of the strength of evidence. So Gerry is comfortable with the strength of evidence that pulse ox adds significantly to usual care. And if I come to a different decision, it's because I look at the same body of evidence and come to a different decision. And I think that's important to recognize.

The last thing I want to point out is that there are many reasons to move beyond evidence in making a recommendation. Clinicians do it all

the time every day in every exam room that they work in. And I think that I don't actually have much problem with that because if we could run the whole world with guidelines, we probably wouldn't need as many physicians or other clinicians. But I think we need to recognize when we do step away from our own rules of evidence and just be honest about it and say we think there's a compelling case to be made for this even though it falls short of our usual rules of evidence. And I just want to say I don't actually have a problem with making recommendations like that, but I would prefer that we be honest with ourselves when we do it.

That's it.

CHAIRMAN HOWELL: Thank you, Ned.

Denise, you had your hand up?

DR. DOUGHERTY: Well, I'd like a little more understanding of the understanding of the following activities taking place and the likelihood of that because to get something funded can take a while.

The other thing is this first bullet -- I don't think it's sufficient really to just study the health outcomes, but also given the questions that are involved, we need to measure the processes of care and probably the instruments that are being used to see if there's a connection between the screening, the processes of care afterwards, and the outcome. Otherwise you won't know the outcome.

The surveillance piece should give you the outcomes.

DR. BOYLE: Well, only if it's linked to infant mortality. So I would add that it's not just monitoring the disease. It's disease that's linked to infant mortality.

CHAIRMAN HOWELL: Coleen, I couldn't hear you clearly.

DR. BOYLE: Oh, I'm sorry. I would link -- it says the Centers for Disease Control shall fund surveillance activities to monitor disease and the outcome linked to infant mortality. It's not just monitoring children who have congenital heart

disease, but it's trying to use that system to see whether or not mortality is impacted by implementation of the screening.

CHAIRMAN HOWELL: Denise, I'm not sure I can answer your question, but we made a series of similar type recommendations for SCID, as you probably remember. And interestingly enough, the letter that came back from Secretary Sebelius, as you recall, said that she accepted that, and she listed the things we needed to do and that we would respond to her in May of 2011 with a report. Now, interestingly enough, those things, actually since our meeting, have been funded and they're actually underway. I mean, we can't predict that this will happen that way, but with SCID, they really have happened.

DR. DOUGHERTY: So if they don't happen, do we withdraw the recommendation?

CHAIRMAN HOWELL: Well, the bottom line is that the Secretary will likely -- if we send this along, she will likely say that she would

expect a report from this committee by a certain date, and there will be a specific date. And I would think that if we're not complying with that, I think that would be a problem. I think that we would be committed to comply with these things.

DR. DOUGHERTY: And about my other comment about needing not only to track health outcomes but to track the processes of care that got to those outcomes.

DR. LLOYD-PURYEAR: I'm sorry. Could you be more precise?

DR. DOUGHERTY: Research activities to determine the care provided and the health outcomes of that care of affected newborns with CCHD.

DR. LLOYD-PURYEAR: To determine --

DR. DOUGHERTY: The care provided and the health outcomes of affected newborns.

CHAIRMAN HOWELL: Mike?

DR. SKEELS: I think everyone else has left me behind, but I need to say this at the risk of sounding like an obstructionist. We spent about

a year hammering out the framework for making recommendations to the Secretary, and I guess my question for you is, are we in that framework or are we coming up with something completely different here? Because it sounds to me like -- Gerry said we're in the zone between 1 and 2. And if we want to change our framework, I think we should do that to adapt. If I'm the Secretary and I read this, I'm going to think -- I don't know what Kathleen Sebelius would think, but I would think are they recommending this or not.

I think we need to go back to our agreed-upon framework to decide which category it fits in rather than suddenly, after seeing the data for the first time this morning, because we didn't even have it to read ahead of time, we're being asked to move forward and wordsmith something that deviates from our agreed-upon practice.

DR. LLOYD-PURYEAR: You did have it. You were sent this two weeks ago.

DR. SKEELS: Well, we had that but we

didn’t have the presentation. You're right. We did have it.

So I guess I just need to know, Rod, do you think that this is -- are we functioning within the framework that we agreed upon in this committee? If so, I’ll shut up.

CHAIRMAN HOWELL: We're functioning exactly as we did with SCID. Let’s put it that way.

DR. SKEELS: I’m not sure that’s true because we used the framework and we said where are we in the framework. And we did make some other recommendations, but we did come down on the side of, yes, we are recommending but without all these qualifications I think.

CHAIRMAN HOWELL: Ned?

DR. CALONGE: I just wanted to answer Mike’s question. So as one of the multiple drafters of the original rules, we had actually had a category that would have fit this better I think that we decided not to use, and it was kind of a

provisional category that we're going to go ahead and add it, and then we're going to watch it. It really set up those times where there's a compelling contextual case. It met the evidence needs for most of the members but probably fell short of the traditional rules of evidence. And so we were so compelled we felt we should add it, but on the other hand, we ought to watch and see what happens. So that kind of phase IV approach. But we consciously decided not to do that.

I think it was a valid, potential category, and I would point out that all evidence-based methods are meant to evolve, that the Services Task Force seems to change theirs about every three years to modify them to make them better. EGAP is facing some of their own methodologic issues of gaps. Anyone on our expert committee could be charged with relooking at phase IV supported addition because I do have concerns we're going to face these issues that don’t quite meet the usual rules of evidence, and yet people

are feeling that the case has been made compelling enough for them to move forward.

CHAIRMAN HOWELL: I would say that Ned has kind of put in words kind of where I am with the subject very well.

Gerry?

DR. VOCKLEY: I think the reality is that nothing that we're going to deliberate on in the foreseeable future anyway will hit category 1 unequivocally based on the kind of rigorous evidence that we would all like to see, and this is just a reality of the diseases. Now, I honestly don’t remember why we got rid of that provisional category, but it does certainly seem like it -- the last two discussions, we've come pretty close to wanting it. And so I would recommend going back to that.

But I also will remind both Ned and Mike that the SCID recommendation had absolutely no details on process or method, and it did come with three bullets that were the starting point for the

bullets up there. So this is very closely modeled after that.

To come back to Mike's specific question, are we recommending this or not, the answer is yes, but we want to have some phase IV monitoring. That's the intent here, and maybe we can make it sound a little bit stronger.

DR. LLOYD-PURYEAR: Could I read what I tried to capture, one, what Ned has said and then we can put it up on the thing. Although there are recognizable evidence gaps, there are compelling reasons for recommending screening for newborns for critical congenital cyanotic heart disease. The addition of pulse oximetry screening for CCCHD to the uniform panel with the understanding -- therefore, the committee recommends the addition of pulse oximetry screening for CCHD to the uniform panel with the understanding that the following activities will also take place in a timely manner and go on with Denise's addition that the NIH shall fund research activities to determine the care

provided and the health outcomes of affected newborns with CCCHD as a result of prospective newborn screening. CDC shall fund surveillance activities to monitor disease linked -- I don't understand this, but disease linked to infant mortality. Is that --

DR. BOYLE: That's fine.

DR. LLOYD-PURYEAR: Okay.

HRSA shall guide State health departments in integration of CCHD screening into their programs. HRSA shall fund the development of, in collaboration with public health and health care professional organizations and families, appropriate education and training materials for families and public health and health care professionals relevant to the screening and treatment of CCCHD.

CHAIRMAN HOWELL: Ned?

DR. CALONGE: Well, I love how we are getting more specific. Thanks, Michele.

I still come back to the issue that I

don't know what -- I think part of this at some point has to be we recommend testing these sites with these probes with this equipment at this time with these cutoffs. And somehow that has to be developed because otherwise I'm not exactly sure what it is we're recommending. So some kind of additional wording that says before this goes to the Secretary, that we have some kind of evidence-based -- what it is that we're screening.

MS. MONACO: Isn't there a recommendation already by the American Heart Association as to what the best time to do the screening is? Is there anything like that?

DR. DOUGHERTY: After 24 hours.

MS. MONACO: Is it all right to put that in there?

DR. CALONGE: I would just be really nervous saying do the screening but we don't know how to tell you how to do it.

CHAIRMAN HOWELL: Chris?

DR. KUS: I kind of share Ned's part.

When you looked at the evidence there -- when are you doing the screening? If it's after 24 hours or some kind of parameters about it because you had the 4 hours, you had the mix of ones. And I would agree that that I'm not clear. If you just say pulse oximetry, that's not specific enough.

DR. CALONGE: Even if we referred them to some external group and said, in compliance with the American College of Cardiology or somebody or pediatric cardiologists -- I just think we have to tell them exactly what it is we're recommending because I just saw a huge variability in sensitivity and only a little bit of danger of specificity if you did it too soon.

CHAIRMAN HOWELL: Tracy and Jane?

DR. TROTTER: Let me ask a question. The only thing since I've been on the committee that was passed has been SCID. I know we heard a lot about technique, but I don't think we said anything about technique. And I presume the States will do whatever they're going to do. I mean, I understand

the concept of trying to make this as specific as possible.

CHAIRMAN HOWELL: The committee has never really weighed in about cutoffs and specific technologies that have been used in newborn screening.

DR. CALONGE: But I think there's a definite way to do this wrong, and that makes me uncomfortable, Rod.

CHAIRMAN HOWELL: Yes, I hear what you're saying.

DR. CALONGE: I mean, the SCID issue -- it seemed like everyone was going to end up doing the same thing, and I've heard the standardization and the standardization is based on something different than this, which is a lab test. Admittedly TMS is pattern recognition and a lot of other things, but I think in general lab folks get to say that this is this condition and this isn’t. I’m just nervous that we're not actually telling people what we're recommending.

CHAIRMAN HOWELL: Mike?

DR. WATSON: We have done the reverse though in looking at Pompe to say that we didn't like Taiwan's fluorometry approach because of the sensitivity issues and wanted to test tandem mass spec in the U.S. population. So it's not like we've ignored it. There was only one assay available here, but there will be -- I mean, every time we look at the LSDs, there are three competing technologies now.

DR. BOYLE: I see Jeff's hand, so I'll let you go first, Jeff.

DR. BOTKIN: Thanks. I was going to say perhaps it would be appropriate to add under the third bullet that HRSA will guide State health departments in the screening standards and integration into the programs. HRSA can then help invite experts in the field to say what's the best way to introduce this technology.

DR. LLOYD-PURYEAR: So HRSA shall guide the development of screening standards?

CHAIRMAN HOWELL: Infrastructure perhaps needed for a public health approach for a point of service, and basically HRSA could convene experts in neonatology, experts in cardiology, et cetera and provide guidance about the technologies required for a public health service. We could put that in there. We could put the requirements needed for a public health approach to point of service newborn screening for critical congenital cyanotic heart disease. And that way you could then come up with whether or not you put the probe on the ear or the toe and what company you use and things of that nature and what cutoffs and so forth. So if we put that in there, that would help with that.

Would that make you more comfortable? Okay. So we're going to add HRSA will work on gathering a group together for infrastructure requirements needed for a public health approach to point-of-service newborn screening.

DR. LLOYD-PURYEAR: So HRSA shall guide

the development of screening standards and infrastructure needed for the implementation of a public health approach to --

CHAIRMAN HOWELL: To point-of-service newborn screening for critical congenital heart disease. That will help take care of that in that area.

DR. BOTKIN: Michele, can we get that up on the screen?

DR. LLOYD-PURYEAR: Yes.

CHAIRMAN HOWELL: Can she email it?

DR. VOCKLEY: I just copied it onto a USB stick.

CHAIRMAN HOWELL: Well, maybe while we're doing that, we could hear from Kof.

DR. OHENE-FREMPONG: Maybe part of the reason why we're not so sure, Ned, is that we've been talking about pulse oximetry, which is actually a method of assessing oxygen saturation. And maybe what we're looking for is low oxygen saturation by whatever the technology is to

determine that. The pulse oximetry is actually quite specific. It's not the only way to assess it. Is a cardiologist in here? Maybe a broader definition that has more of a clinical understanding and not so much the method which would then force us to try to specify exactly how the particular method --

DR. CALONGE: Kof, the other thing that leads to my discomfort were phrases like everyone knows that if it's under 90, you need to be treated, but that's not the cutoff I heard used by any method. It was 96 or --

CHAIRMAN HOWELL: But if we put this wording in there, hopefully that will get that --

DR. CALONGE: I agree. I think having someone look at it and come up with something.

CHAIRMAN HOWELL: Dr. Govindaswami is here. Roger, if he could comment briefly on some of the conundrums that we're dealing with.

DR. GOVINDASWAMI: Thank you. Just a few comments.

I think transcutaneous pulse oximetry is a simple way you can do co-oximetry in the blood stream, but that’s fraught with more problems and the baby screams. There are more shunts. So we wouldn't recommend any way other than transcutaneous pulse oximetry for screening in the context of these discussions.

DR. OHENE-FREMPONG: My point is we screen for something, not use this method. I mean, if wanted to find that a particular enzyme is low, that’s what we're looking for, and we're not asking people to use a method.

DR. GOVINDASWAMI: Correct.

DR. OHENE-FREMPONG: Can we use a term that actually defines what abnormality we're looking for? In this case, if it is O2 desaturation --

DR. GOVINDASWAMI: Yes, I think that's correct. I think if you said screening for desaturations in babies, or whatever language you use -- I get the point.

But I think the cutoffs is not the big issue because most of the studies look at that 95 as the cutoff. The issue of the evidence gap of what happens if we don't do this I think is addressed in the large Swedish study where the babies who were screened and picked up were much less likely to die I think. I don't have my notes with me, but it's something like 60 versus 5 of the 100 who got picked up who didn't get screened and then subsequently died. So there is a cost of life.

And I like the addendum of the infant mortality connection because I think that will be an easy way to make it.

But I'm just very encouraged by these discussions and the points raised on the specific issue of separating moms and babies to do pulse oximetry. The comment I have is, you know, when we do the hearing screen, it's a much more prolonged test and we take them to a quiet room. This testing takes about 2 minutes, and even at our

center where we've done 4,000 babies using the two-site technique, which I think is what we do, at our center it works best for us. There's not a time constraint. We don't even have to separate them from the mom. There are people looking at can we do it in the mom's arms. So I think those are technicalities that we can overcome.

I think the very real issue of how will it be implemented in different sites -- some of the questions you are discussing are things I've certainly agitated over because the way I do it at my medical center where I have pediatric cardiologists and echotechs and everything 24/7 may not be the way I'm going to implement this in Gilroy, which is 25 miles away. I may recommend doing the studies a little earlier knowing that I have a higher false positive rate, but that way somebody on call will hear about this baby because if you fail the earlier screen, the only thing you do is repeat the screen. You don't run away and do an echocardiogram. So you add a $5 to $10 cost as

opposed to a $500 or $1,000 cost. So there might be specific instances where regional programs can decide how they do it best within their systems.

CHAIRMAN HOWELL: Thank you very much.

I think that this discussion has been helpful because, number one, we've taken pulse oximetry out of the recommendation because we really are screening for critical congenital heart disease. That, obviously, was very helpful, Kof, to bring that up.

And if we add then the fact that HRSA will oversee an effort to work on this in a public health arena, then we'll get around that.

Chris?

DR. KUS: I guess I'm missing something because I thought part of it is that there is a good screening way of doing the screening, and if you take the pulse oximetry out, it seems to me that you're asking them to come up with that. I think they proposed here that we've got screening programs that you'd use pulse oximetry.

CHAIRMAN HOWELL: We've recommended screening for PKU and we certainly don't say you need to use tandem mass spec.

DR. KUS: Yes, but PKU was before this body.

CHAIRMAN HOWELL: Well, the thing is that I think that we've heard that although everybody is using pulse oximetry, there may be a better way that evolves, and it would be nice not to have that in our recommendation.

Does everybody want to put pulse oximetry back?

DR. VOCKLEY: I don't see any reason to. We recommended SCID screening without mentioning TRECs, without mentioning --

DR. DOUGHERTY: Question: What was the evidence review on? Was it looking at pulse ox? Pulse ox. So the evidence review was not about using other techniques.

CHAIRMAN HOWELL: Roger?

DR. DOUGHERTY: But the evidence review

was not about those things.

ROGER: I would really be very wary of putting methodologies into your recommendations because although today there may be one thing that's the best, who knows about tomorrow? And if you tie a recommendation with methodologies and then next year something comes up that's 50 percent better, that could just inhibit progress. I think that your job is -- it's very nice that you're taking the comprehensive responsibility of all these factors, but it's kind of micromanagement.

DR. DOUGHERTY: But then could we change that NIH recommendation so that it also tracks what kind of method was used to do the screening? You know, sometimes techniques change. They become the hot, new thing, and they're not.

CHAIRMAN HOWELL: I think that hopefully Alena is going to have this up on the screen very soon. There it is.

Would our nominator like to read that for us?

DR. VOCKLEY: I can. Although there are recognizable evidence gaps, there are compelling reasons for recommending screening newborns for critical congenital cyanotic heart disease. The committee recommends the addition of screening with the understanding that -- the first bullet -- the National Institutes of Health shall fund research activities to determine the care provided and the health outcomes of affected newborns with CCCHD as a result of prospective newborn screening; that the CDC and Prevention shall fund surveillance activities to monitor disease linked to infant mortality. The third bullet: The Health Resources and Services Administration shall guide the development of screening standards and infrastructure needed for the implementation of a public health approach to point-of-service screening for CCCHD. And then HRSA shall also fund the development of, in collaboration with public health and health care professional organizations and families, appropriate education and training

materials for families, public health and health care professionals relevant to the screening and treatment of CCCHD.

CHAIRMAN HOWELL: Comments on that, Jeff?

DR. BOTKIN: No.

CHAIRMAN HOWELL: This is a modification of your original recommendation.

Is there further discussion of this recommendation at this point in time? Ned?

(Laughter.)

DR. CALONGE: I think you've spun it as well as it can be spun.

(Laughter.)

CHAIRMAN HOWELL: Speaking as a professional spinster.

Is there anybody else who would like to comment about this recommendation? Jane?

DR. GETCHELL: I think this has really come a long way since we started, and speaking from a State program perspective, I do appreciate all that has been added to it in terms of HRSA support

technologically, financially, and guidance-wise. That's really very helpful.

The question I have -- and this really pertains not just to this but probably future diseases that we'll be looking at. What are the implications of adding it to the uniform screening panel? Does that automatically mean that it becomes the responsibility of State programs?

CHAIRMAN HOWELL: It's a recommendation to the Secretary. I don't think it automatically becomes a requirement of the State. I think many States will obviously adopt it into their programs.

DR. DOUGHERTY: It will be expected to be part of long-term follow-up at the State level.

DR. GETCHELL: Yes, it will.

DR. BOYLE: But that's an easy one. They already have their birth defects monitoring programs linked with infant mortality, and they can do linkages to hospital discharge. They can do lots of stuff there that's already in place.

DR. GETCHELL: It's already in place but,

for example, with this one it will require some infrastructure development, new expertise, additional staff, those kinds of things. And I think this recognizes that.

DR. BOYLE: Again, I would encourage them to use their birth defects programs. This is a wonderful reason for them to exist. It gives them a reason to link with outcomes for children rather than just monitoring for birth defects.

DR. KUS: I mean, I share the idea that as a State you want to implement which things that you think are recommended, and to implement this, we always come down to the idea of having the resources to be able to do it and who does the resources. And I think we've talked a lot about the partnership between States and the federal government. I would say, listening to Coleen about our birth defects, our birth defects wouldn't be able to do this right now without added resources, and it’s not the system that we would probably use to do that.

DR. BOYLE: Again, I think it's an opportunity for those systems to expand.

Can I make one modification to the CDC one, which is to monitor disease linked to infant mortality and other health outcomes?

DR. LLOYD-PURYEAR: Oh, good.

DR. KUS: I guess the idea is expansion takes resources, and I think that's the concept that we're really struggling with. You want to do this and how do you do it?

CHAIRMAN HOWELL: Mike?

DR. WATSON: I think the problem is in the actual Newborn Screening Saves Lives Act which I think pretty clearly says that if a State does not meet the standards established by this committee, there could be an impact on their federal funding.

DR. LLOYD-PURYEAR: If a State receives funds under section 1109 of the Newborn Screening Saves Lives Act, they have to agree to be in the -- either having adopted this committee's

recommendations or be in the process of adopting. But that's the only requirement. Well, very few States --

DR. WATSON: Get money through that pathway.

DR. LLOYD-PURYEAR: Well, until we have more money appropriated, that's really not an issue.

(Laughter.)

DR. WATSON: It's certainly never been used, but it is sort of, I think, the big dog.

CHAIRMAN HOWELL: It might be substantial going forth I think is what Michele is saying, if more money flows in.

Is there anything new or something to say about this?

DR. DOUGHERTY: Yes.

CHAIRMAN HOWELL: Denise has something I can tell.

DR. DOUGHERTY: I'm making a friendly amendment to my own words. So what I'm really

trying to get at is the NIH shall fund research activities to determine the relationships among the screening technology, the diagnostic process, and the care provided, and the health outcomes.

CHAIRMAN HOWELL: Any further comments about this recommendation? Mike, you look like you have something to say.

DR. SKEELS: No, other than I have to leave for the airport and I'd like to leave the committee on a yes vote.

(Laughter.)

CHAIRMAN HOWELL: Yom Kippur is rapidly closing in on us so that we need to get this settled and go deal with even more important events of the evening.

I can call for a vote, but I want to be sure everybody has their word before we vote.

Those favoring this recommendation? Denise, do you still have something to say?

DR. DOUGHERTY: She's asking me for what I said.

CHAIRMAN HOWELL: I see. And does she have it?

DR. DOUGHERTY: Yes.

CHAIRMAN HOWELL: So we've got that.

So we're going to take a vote. Those favoring this nomination that's been made by Dr. Vockley and seconded by Dr. Botkin and discussed exhaustively by this group, raise your hand.

(A show of hands.)

CHAIRMAN HOWELL: Peter, is your hand up? Okay, thank you very much.

Those opposing this nomination?

(A show of hands.)

CHAIRMAN HOWELL: We have one person opposing.

Is there any abstention?

DR. LLOYD-PURYEAR: Dr. Guttmacher is absent.

CHAIRMAN HOWELL: He had to leave for an appointment, so he's absent.

That is it. It passes overwhelmingly.

So thank you very much. I think that was a very worthwhile discussion.

I hope that there's not a lot of other committee discussion.

DR. GELESKE: Can I just ask? Can you email that out right away? Because I'm sure the AAP will be very interested in that recommendation and I want to get the wording correct.

CHAIRMAN HOWELL: I'm sure that it will be available. It can be emailed promptly.

So is there any other committee business outstanding?

Let me bring out that we really now would like to have material for the January meeting.

DR. LLOYD-PURYEAR: Yes, we do. We do have committee business, the HIT Workgroup. And Sharon had to leave go to her sabbatical.

CHAIRMAN HOWELL: She has left for England.

While we're waiting for the slide to arrive, you're going to get an email from Altarum.

That’s the company that organizes this group. Please fill out the survey about the meeting and how it worked for you as far as the facilities are concerned.

We would like agenda items for January. Please send them to Michele so that we’ll have those to discuss.

The meeting dates for next year are May 5th and 6th, September 22nd and 23rd, 2011. We, hopefully, will not be in the midst of some major holiday in January we hope. We do have the dates, but I don't have the dates.

DR. LLOYD-PURYEAR: That’s January 28th and 29th.

CHAIRMAN HOWELL: So this is the recommendation that --

DR. LLOYD-PURYEAR: If you go to the second slide, yes.

CHAIRMAN HOWELL: Fundamentally, as you recall, this was a recommendation that this committee support the quality measures that would

be looked at as proposed by these groups.

DR. DOUGHERTY: Michele, you emailed sort of my version.

DR. LLOYD-PURYEAR: This is what Sharon came up with. Sharon and Alan.

DR. DOUGHERTY: You weren't asking other people to weigh in?

DR. LLOYD-PURYEAR: I did it to the whole group, and Sharon left and it has this, part two. These are just friendly amendments.

CHAIRMAN HOWELL: Is the thing that we're being asked to vote on, this particular slide?

DR. LLOYD-PURYEAR: Yes.

CHAIRMAN HOWELL: Read this and see what you think of that. Denise, does this capture your --

DR. DOUGHERTY: No. That language about communication processes is puzzling to me.

DR. LLOYD-PURYEAR: That is what the quality measures are for.

DR. KUS: The "such as" -- I thought we

were initially proposing that the measures that they proposed were ones we wanted to say go forward with. “Such as” is still pretty weak to me.

DR. DOUGHERTY: And this one doesn’t say as long as the measures meet the NQF criteria for scientific --

DR. LLOYD-PURYEAR: Yes, it does. We don’t that they don’t meet the scientific acceptability. We recommend that they accept the scientific acceptability.

DR. DOUGHERTY: But I thought our point earlier was we only endorsed them if they meet the NQF criteria. So to recommend that NQF is going to assess it doesn’t make any sense because NQF is going to assess it. That’s what they do.

Ned, can you help?

DR. CALONGE: I actually think this -- I mean, other than the communication, I don't know if it’s communication or follow-up or care coordination or something. Communication is part of it. I actually think the overall structure is

okay. It says we support the endorsement of newborn screening quality measures. So that's good.

CHAIRMAN HOWELL: That's good.

DR. CALONGE: That's an important first statement.

The last one is that they should assess, you know, what we talked about, the validity and -- it's feasibility as well as scientific acceptability. Can we actually get those?

But I think that last sentence captures that. We can't say we endorse these measures because we don't know the scientific validity and the availability --

CHAIRMAN HOWELL: But we've asked that they look at that.

DR. CALONGE: And they look at it. So I think those two are fine.

My only problem is with is it just communication process or measures assessing the --

CHAIRMAN HOWELL: Why don't we just take

that middle thing out? Alena, why don't you just take that out. Just take that out.

This is very straightforward. I hope no one around the table --

DR. LLOYD-PURYEAR: Do you want us to add feasibility?

DR. CALONGE: Yes. I think feasibility is always important.

DR. LLOYD-PURYEAR: Can you add we recommend NQF assess the scientific acceptability and feasibility?

DR. BOYLE: I would get rid of everything after the "such as."

CHAIRMAN HOWELL: What did you just say?

DR. BOYLE: I would just do what Ned said, put a period after "quality measures." Period.

DR. VOCKLEY: Why not just eliminate the "such as" part and say screening measures proposed by?

DR. BOYLE: Just put a period there.

DR. CALONGE: I think there actually are some that have been recommended. I actually kind of like that.

DR. DOUGHERTY: Yes, I don't like that either because that’s what we wanted to get away from.

DR. CALONGE: Those are specific. So we're actually making a specific recommendation by including “such as those.”

DR. WATSON: Or just say “those proposed by.”

CHAIRMAN HOWELL: But those are the ones we actually -- I mean, that’s very straightforward. We certainly would like the newborn screening thing, and we would like to be certain these are reasonable things to be doing. Isn’t that what we say?

DR. CALONGE: Something like “as proposed.”

CHAIRMAN HOWELL: Any further comments about this?

Those favoring this motion, please raise your hand.

(A show of hands.)

CHAIRMAN HOWELL: Is anybody abstaining?

(A show of hands.)

CHAIRMAN HOWELL: Are you abstaining? So we have one abstention.

So it passes unanimously. So thank you very much.

DR. DOUGHERTY: I'm voting no.

CHAIRMAN HOWELL: You're voting no, okay.

DR. DOUGHERTY: I think it's different from what we were trying to do.

CHAIRMAN HOWELL: Thank you very much. Ladies and gentlemen, I think that this has been an extremely productive meeting. Lots have been done and a lot of progress and so forth. And I thank you. And I wish you a very successful holiday, and we will see you in January.

But we need a motion to adjourn.

DR. VOCKLEY: So moved.

CHAIRMAN HOWELL: I think Denise is going to oppose that.

(Laughter.)

DR. DOUGHERTY: No, absolutely not.

CHAIRMAN HOWELL: I see a unanimous vote to adjourn. Thank you very much.

(Whereupon, at 2:50 p.m., the meeting was adjourned.)