SECRETARY'S ADVISORY COMMITTEE ON

HERITABLE DISORDERS IN NEWBORNS AND CHILDREN

Friday, May 18, 2012

MORNING SESSION

8:30 a.m. — 12:00 p.m.

Hilton Alexandria Old Town Hotel

1767 King Street

Alexandria, Virginia 22314

APPEARANCES

COMMITTEE MEMBERS:

JOSEPH A. BOCCHINI, JR., M.D. - Chairman

DON BAILEY, PH.D., M.Ed

CHARLES HOMER, M.D., M.P.H.

STEPHEN MCDONOUGH, M.D.

DIETRICH MATERN, M.D.

ALEXIS THOMPSON, M.D.

ANDREA WILLIAMSON, B.A.

EX-OFFICIO MEMBERS:

COLEEN BOYLE, PH.D., M.S.

SARA COPELAND, M.D.

DENISE DOUGHERTY, PH.D.

KELLIE KELM, PH.D.

MICHAEL LU, M.D., M.P.H

MELISSA PARISI, M.D.

REPRESENTATIVES

NATASHA BONHOMME, B.A.

FREDERICK CHEN, M.D., M.PH., FAAFP

REPRESENTATIVES (continued)

JANE GETCHELL, DR.PH., MT (ASCP)

CAROL GREENE, M.D.

BENNETT LAVENSTEIN, M.D.

NANCY ROSE, M.D.

BETH TARINI, M.D., M.S., FAAP

EMIL WIGODE

MARY WILLIS, M.D., PH.D.

C O N T E N T S

P R O C E E D I N G S

CHAIRMAN BOCCHINI: Well, good morning, and welcome to day two of the Secretary's 27th meeting. I hope everyone enjoyed the evening last night.

And to start today, we need to take roll again. So I will go down the list, and if you'll just answer here.

Don Bailey?

DR. BAILEY: Here.

CHAIRMAN BOCCHINI: I am here. Coleen Boyle?

DR. BOYLE: I'm here, too.

CHAIRMAN BOCCHINI: Sara Copeland?

DR. COPELAND: Here.

CHAIRMAN BOCCHINI: Denise Dougherty?

DR. DOUGHERTY: Here.

CHAIRMAN BOCCHINI: Charles Homer?

DR. HOMER: Here.

CHAIRMAN BOCCHINI: Kellie Kelm?

DR. KELM: Here.

CHAIRMAN BOCCHINI: I know Michael is not

here yet. Stephen McDonough?

DR. MCDONOUGH: Aye.

CHAIRMAN BOCCHINI: Dietrich Matern?

DR. PARISI: Here.

CHAIRMAN BOCCHINI: Melissa Parisi?

DR. PARISI: Here.

CHAIRMAN BOCCHINI: Alexis Thompson? Not here yet. And Andrea Williams?

MS. WILLIAMS: I'm here.

CHAIRMAN BOCCHINI: And then from the representative members, Beth Tarini? Not yet? Okay.

And Nancy Rose?

DR. ROSE: Here.

CHAIRMAN BOCCHINI: Jane Getchell?

DR. GETCHELL: Here.

CHAIRMAN BOCCHINI: Chris is not here today.

Bennett Lavenstein?

(No response.)

CHAIRMAN BOCCHINI: Mary Willis?

DR. WILLIS: Here.

CHAIRMAN BOCCHINI: Natasha Bonhomme?

MS. BONHOMME: Here.

CHAIRMAN BOCCHINI: Emil Wigode? Am I messing your name each day?

MR. WIGODE: Emil.

CHAIRMAN BOCCHINI: Emil, okay. Thank you.

And Carol Greene? Okay.

DR. CHEN: Joe, this is Freddie Chen. I'm on the phone. Can you hear me?

CHAIRMAN BOCCHINI: Hey, Fred -- Freddie. Yes, we can hear you. Welcome.

DR. CHEN: Good morning.

CHAIRMAN BOCCHINI: Good morning. Where are you, Freddie?

DR. CHEN: I'm in Seattle, but I'm up and ready to go.

(Laughter.)

CHAIRMAN BOCCHINI: Okay. All right. Well, we expect that you'll be vigorously involved in all conversations then. Good.

So this morning we're going to start off with subcommittee reports. But our goal today is

not only to hear the subcommittee reports, and it's very clear the subcommittees have been working very diligently to organize their priorities and then pick projects to be involved in. But today in addition to hearing where they are, after yesterday's summary and opening session and then their subcommittee meetings, this committee will now review their projects and the priorities that they've set, and then help hone down to the key projects and prioritize with them how they will each proceed and which projects they will pursue.

So what I thought we would do is rather than wait for each subcommittee report to discuss the prioritization at the end, we'll discuss that after each committee report.

So we're going to start off with the Subcommittee on Laboratory Standards and Procedures, and Dieter, who will make that presentation of the subcommittee's work yesterday. So, Dieter?

DR. MATERN: Good morning, everyone. So we met yesterday. We were very efficient, done at 4:00, to discuss the following issues.

So here you see the members of the committee. Fred unfortunately could not attend, so I am presenting on his behalf. And on behalf of the committee -- and you see at the bottom our agenda, which was primarily the discussion of the priorities and projects as was just mentioned. And then we had Alex Kemper come in and talk to us about the review of conditional review process report.

So we looked -- we have three priorities identified. The first one is review of new enabling or disruptive technologies, and among those, we have three projects. The first one as we have it right now, but I guess we'll discuss, is that we feel it might be helpful if we were to take a look at the pros and cons, which includes any uncertainties about all kinds of platforms, technological platforms, used in newborn screening, whether they are old, so meaning they're currently used, or coming potentially into laboratories for the nominated or current conditions on the uniform panel. And that the goal is here to help States make to make informed decisions on what should they

do as they add new conditions or see how they can make sure that they capture all the conditions that are currently on the panel.

So if we think about an old platform, it would be tandem mass spectrometry. And we look at succinylacetone, which apparently is still not measured in every screening laboratory as part of the first tier screen, along with the amino acids and acylcarnitines.

So this would be one example that we already discussed last time, how can you -- and actually in September already it came up, how we can help all screening laboratories to consider which method they will use to capture succinylacetone. And there are probably three or four different approaches at this point.

The second project is also meant to help screening programs in their implementation of new conditions that have been added to the uniform panel. So an example here would be SCID that apparently is recommended, but it's not implemented in every State. There are two or three different

approaches on how you do the screening assay, although every screening assay measures the same analyte. So the screening labs I guess are struggling a little bit as to which approach they should take. So this is, again, something where our committee might be able to help.

One could provide potentially a repository of the SOPs. I think the CDC recently published or has a method that they implemented in their labs, so I'm sure they would be willing to share that very freely.

Other conditions related to newborn screening, most screening laboratories are, I think, willing to share their SOPs so that others can take a look and decide for themselves what would work in their laboratory.

And the other goal would be to provide a slide set that screening laboratories could use to educate the decision makers in their States on how to -- on why a new condition that has been recommended should really be implemented in their State as well.

So a slide set could actually be a true slide set, a Power Point presentation that provides information about the disease, about the test, about the cost, about the implications for the screening laboratory so that they can take it to their advisory boards, their administrators, to the legislature, whoever may need it, maybe the media as well. And some States might need some education so that they portray newborn screening as what it is in and not what they think it is.

The third project might be the easiest one to accomplish. HRSA has funded a total of six years the region 4 tandem mass spec data project, which is basically run by Piero Naldo. This is coming to a close at the end of this month. And so it might be an idea to have Piero come back to the committee and give a summary of what he's done all these years, present the data that were collected and the tools that were developed as part of the project.

I think it would be important to look at the potential impact that the program -- what they have done -- has had on screening programs that use

the tools and the data and see whether it made any difference.

And then something that we didn't discuss, but I thought to mention also is since Piero gave a training course, it was a week long. Anybody here who has participated in this may have their own ideas, but I would think that all of them would agree that it was a very unique experience to be locked up with Piero for a week --

(Laughter.)

DR. MATERN: And realize that he's not such a bad, and actually likes to share his knowledge. So maybe there's some value in using his syllabus or whatever to continue the training course in one way or the other, that goes to that level of detail.

The second priority we have at this point is to provide guidance for State newborn screening programs in making decisions about implementation, integration, follow-up, and quality assurance. There's clearly some overlap to the first priority.

The first project here would be to compare

performance metrics as they exist right now, or as they are developed, so looking at the APHL quality indicator metrics that quite frankly I don't really know too much about. So it's probably going to get to look at them.

But also to review the newborn screening case definition that we heard yesterday about which, I think, are clearly important so that we all know what we're talking about when we talk about a disease and cases.

The second project, what about point of care newborn screening. The idea was here to kind of lay out for the committee the landscape of what is going on right now in public health programs as to how it is dealt with, with one example. The easiest one, I guess, where most of the information would be available would be the screening for hearing loss, who is doing what in the hospitals, who is doing what in the State labs, how is this done? How is information getting back to the programs? How is it ensured that every baby is actually having a screen, and then is not lost to

follow-up. And if we were to identify a perfect model, then that might be something that everybody should know about so that other programs could emulate those.

And a third project would be to develop a tool to capture delayed or missed diagnoses. And this is something that Harry Hannan apparently has a great interest in. I think it is important, and the subcommittee agreed with that, that it would be nice to know whether there are two false negatives or false negatives that could be avoided. But otherwise how does a screening program actually find out about whether there is a missed case?

So we had a short discussion about how this could be done. Don't really have a good idea yet how it could be accomplished.

If the medical record -- the electronic medical record may be able to do this, my idea would be you just put a little program in there that whenever you choose a condition that is part of the newborn screening program, it flags and it asks you why the heck did you put that condition in here

because this condition should be known now already since the second week of life at least. So that would allow you to capture cases that may not have been screened, as long as there's feedback going back to the screening program.

The third priority would be to establish a process for regular review and revision of the uniform panel, and to recommend specific changes to technology when indicated. So that kind of goes back to maybe the succinylacetone story where you have a technology that you use already, but you might have to tweak it a little bit so that you can make sure you can identify all patients.

So one project would be how do you remove a disorder from the uniform panel because there's apparently consensus one has to find through the Evidence Review Group probably or with help from, is which condition actually does not really deserve to be there because it might just be a biochemical variant, but not a true disease, and, therefore, we're not really doing anyone a favor by labeling people with a specific condition.

And then the question is, how do you actually stop screening when you have multi analysis that you're looking at where you might have overlap for the condition that you want to get rid of with one that you want to keep. And I think the conditions that I could think of, that shouldn't be a problem.

And then how can you move a condition from secondary to primary target? I guess we have to look at this if there are any, and then how do you do this? And then finally, it's not really a bullet point here because I didn't know how to put it. But I think there will be basically for this priority an interaction with the Evidence Review Group, but also our subcommittee could provide some technical information to the Evidence Review Group, and they look at new conditions and see how those could be screened for.

And that is all I have.

CHAIRMAN BOCCHINI: Great. That was a very nice summary and very clear that the work of the subcommittee had really evolved to very specific

recommendations.

Are there any questions at this point? Comments?

DR. HOMER: I think it'll become clear as we have the full conversation, but there clearly is significant overlap between what this priority is and the long-term follow-up. So I look forward to the discussion as we -- I don't know if "adjudicate" is the right word, but at least come up with some strategy for being clear we're not duplicating effort.

CHAIRMAN BOCCHINI: I think you're right. I think there clearly is some overlap that's going to occur, and I think that's a good opportunity to try. And think "adjudicate" is not a bad word. That might fit.

So we can go back to the first priority? Let's just kind of go through a couple of things and raise a couple of points about how we might accomplish or what we might need to accomplish some of the things that you've mentioned.

How do you see completing project number

one? Would this be the committee making a decision and bringing it forward to the Secretary, or would you see providing sort of a summary of current data that would then be in the form of a publication?

And I raise that in part to consider, like MMWR, something similar to that, or MMWR as a place where this committee could put things like that that would help make this a more formal recommendation for the States. And for that, it would mean that the CDC might be involved through its laboratory area. And I don't know whether Carla is here at the moment to consider that. But to me, it would be appropriate to have a more formal process where that might be done in that kind of a fashion.

MS. CUTHBERT: Yes, that could be done. I have somebody in my lab already who is considering looking at some of these things so we can work together to actually make that happen.

CHAIRMAN BOCCHINI: Okay, because it seems -- I know we've discussed that before and I think the consensus is very clear that the data supports this, then it would be how should we get this done

most rapidly?

DR. BOYLE: So are you thinking of an R&R report and recommendation?

CHAIRMAN BOCCHINI: Right.

DR. BOYLE: Yeah. So, I mean, there's a different standard now for that, and it really has to be an evidence-based review process. So it would be great. Actually I think that would be a nice way to characterize that.

CHAIRMAN BOCCHINI: So, to me, the way to handle that on a regular basis when these things come up would be to do a special R&R format because there's the weekly report, and then there's these R&T reports that provide a significant amount of evidence, you know, background information and then specific recommendations. And I think if this committee could work closely to do that with the CDC, I think that would be a really good way to get that done. And, to me, that sounds like a top priority because I think it's clear that this committee believes that's the case.

So how would we go about sort of working

with Carla to make that happen, just work directly with you?

MS. CUTHBERT: Yes.

CHAIRMAN BOCCHINI: Okay. Yeah, oh, perfect. Okay, all right. Denise?

DR. DOUGHERTY: It seems we might go through these again with an eye toward what makes sense for the -- I think you're getting to this -- what makes sense for the subcommittee to try to do, and what makes sense as a recommendation for someone else that's more of a resource. Not that any of us have great resources these days, but more resource to try and do some of these things.

Like putting together guidance for all the labs in the country just doesn't seem like something a subcommittee could really do. What?

DR. BOYLE: But Carla --

DR. DOUGHERTY: But Carla could something like that. So, yeah.

CHAIRMAN BOCCHINI: Right. The question is how to then format that if the recommendation from our committee is to consider that, then put it

under the purview of CDC to then use their resources to develop that, yeah. If it's possible, right.

DR. COPELAND: We could also -- I mean, just as the Follow-up and Treatment Subcommittee had developed workgroups, we can also do that with the Lab Standards. And we have a lot of resources in there. We have APHL as a member of our subcommittee. We have CCDC, HRSA.

So I think that we don't necessarily need to place this on any one person, but rather this is a priority, and the subcommittee in turn -- just as the subcommittee can decide the best mechanism to get it, but I hesitate because I don't want you telling HRSA how to spend their funds. And I wouldn't dare to tell ARC how to spend their funds.

And so I think we need to be really, really careful that if we list priorities, and then the subcommittees in turn can determine the best mechanism to get there.

DR. DOUGHERTY: But I guess what Joe is trying to figure out is what's the -- I mean, these are all great things it sounds like. But it sounds

like an enormous amount of work for a subcommittee to take on. These are volunteers. I don't know if anybody else who's led a subcommittee wants to speak on this, but what -- and how much resources HRSA has to -- you know, very often these things turn in to, well, we need an expert meeting to do this.

You know, so just thinking through the resources really carefully and figuring out what's actually doable within what time frame I think is a good idea.

DR. MATERN: I think the time frame is the biggest question. I mean, if we want to do this in two years, all of those things, it's probably not something the committee can do even if it asks their friends to help.

I don’t know if you pass it onto another group that they would be able to do it either because they might not have the background information. In newborn screening, that's sometimes difficult to find.

CHAIRMAN BOCCHINI: Yeah. And I think this is a good example. If the subcommittee has a

good understanding of the changing technology or the changing analytes to look at and those things, and now this has become clear that this is a better way to do the test, then it would mean that the subcommittee should -- we need to figure out how to bring that to the full committee so the full committee could make a recommendation to the two States in some fashion to make a change.

But that's why I think that we need to partnership with those agency or organizations to then have that implemented through publication or recommendation because I think going through the Secretary's office is one way to do it, but then going using the -- yeah, using CDC that has that capacity to get that information out and the relationship with the public health programs, you know, might be a good way to do that.

So I think we need to flesh that out, and I think you're right. I don't think we need to try and control what other organizations do. But I think we should have a partnership with them in a way to define what's the best way to get that

implemented in the laboratories. But to me, that's a primary project that looks like it would have good, immediate benefit if we could get that out.

But I see it coming -- perhaps the subcommittee could ask for additional help from others outside of the subcommittee as well as the rest of the committee, and we could bring that as a project to the committee for its final decision. So I think we just need to make that more of a project that we want to get done and figure out how to do that together. And I think CDC is probably the best partner for this one.

And then the second implementation tool for new conditions, I think that's an excellent idea. I think that what was done with SCID was really very -- again, working with CDC, having CDC partner with public laboratories, and having that meeting to bring all the individuals from the laboratories in to learn the technique, to watch what CDC was doing. I think that was a very good way to do that. And so essentially, that was done very nicely for SCID, and I think that's a good idea

to do for each of the subsequent things that we consider for the RUSP. So I think that should be done with each one that we do, and so that could be part of what we do. So that's not something we need to do all the time, but that would be something that could be done with each condition that's being evaluated or a decision has been made to do.

And, again, I think that would -- again, partnerships would help get that implemented in a more rapid fashion once the Secretary made the decision to add something to the RUSP.

And then project three, I think, again, that's a good role for the subcommittee to be aware of that, and to make the committee aware of when we need to have somebody come and talk about that, and then make a decision whether to endorse that or support that. And, again, that might lead to making decisions with the CDC as to how to put that into States laboratories in the best way.

So I like these three, but I think from my view, project number one is the one that I think probably needs to be addressed most quickly in such

a way to bring that forward out of these three priorities.

Any comments from the committee -- consideration for that? Feedback from Dieter?

DR. MATERN: Still for project one and the pros and cons, we would have to get some guidance as to which platform we should address first because, again, I think the scope would be too large if we tried to address every single platform that's currently used in laboratories.

CHAIRMAN BOCCHINI: Right. No, I agree with you, and I think I'm using the one example, which is the one that we've talked about a couple of times about changing the analyte that's being evaluated would be a specific project rather than trying to look at all of the platforms for nominated and current conditions. I think at this point, it would be looking at the one that you've all indicated needs a change.

Carol?

DR. GREENE: Apologies if this is redundant because I was --

CHAIRMAN BOCCHINI: The microphone?

DR. GREENE: Oh, sorry. Apologies if this is redundant because I was putting together my slides. But I see potential synergy between project proposal number two and one of the ones that the Follow-up and Treatment Committee is going to propose.

Of course we were looking at the follow-up and treatment, but we were also thinking about looking at implementation with an eye to the possibility -- with a need to learn from what's gone before so that when people implement new conditions, they do it with, you know, benefit of the previous wisdom. And this is focused on the lab site, but it could be put together as a package, work with the Education Committee. So the goal would be that when new screening is implemented, that there would be a -- some guidance that would come forward to help people know how to implement not just the lab, but the short-term and the long-term follow-up, what kinds of things need to be in place. I see a lot of potential synergy there.

CHAIRMAN BOCCHINI: Good, because I think that's -- the primary goal here is that even though subcommittees may bring forward some of these ideas, that when something like this happens, we're going to deal with this as a committee. And so bringing those things together, same individuals working together within the overall committee would be the answer. So it would be someone perhaps from -- making sure that we address the issues, and follow up implementation, and laboratory all together, so good.

Additional comments? Okay. Let's go to priority two. So these three, I think clearly project three, as you said, you're in the early stages of discussion. And that might be difficult with the stage of electronic health records right now to do what you suggested, except in a large organization that has that available. And that could be the possibility of developing a pilot project through that.

So comments related to these? Steve?

DR. MCDONOUGH: Did the issue of historic

blood spots come up in the committee as far as any activities for the next year?

DR. MATERN: For next gen sequencing?

DR. MCDONOUGH: Retaining blood spots for research.

CHAIRMAN BOCCHINI: Yeah. So the Secretary currently has the recommendations made from this committee for that. So we're awaiting the response for that. Carol?

DR. GREENE: This also overlaps beautifully with two of our proposals.

CHAIRMAN BOCCHINI: I'm sorry, I missed the whole thing.

DR. GREENE: This also overlaps beautifully with two of our proposals.

CHAIRMAN BOCCHINI: Right. Okay. Yeah. And I think there are a couple of things we can extract from this that are committee issues. And, to me, point of care in newborn screening is a committee issue just like we talked yesterday about ethical issues that might be involved with a variety of different aspects of what we do, that's probably

more of an overriding committee issue.

And so I think our role in looking at the landscape, et cetera, I think that's very important areas, but it might be more that the committee has to deal with that rather than the subcommittee.

So Carole and then Denise?

DR. GREENE: And actually in the State of Maryland, I headed the equivalent of the Lab QA Committee for subcommittee for the implementation -- design implementation of congenital heart disease. And our job was easy because we said it's hospital based. The States can make sure people know how to do it and have guidance. But, you know, there's no lab -- the QA is all at the level of the hospital, does your equipment work.

So I actually think that with due respect and looking forward to working with Lab Committee that project number two at least is really a follow-up and sort of large program. I'm sure there are other ways of looking at it, but I'm just not sure for point of care for hearing, I don't see it as a lab problem. There are going to be plenty of lab

issues that could be point of care, but I don't know that we have one at this moment.

DR. DOUGHERTY: It's lab standards and procedures. And our charge definitely covers parts of the technical aspects of the screen itself. And so as far as purview, I would disagree.

And I think that different States do it so very differently, and that was the point is with hearing screening there's a huge variety in loss to follow-up. You can go from 10 percent to 40 percent that have loss to follow-up.

And as far as I know, and I don't know if Irene or Bonnie can comment on this, that a comparative analysis of the follow-up process has been done to determine if it's a State versus attached to the lab. So that's the whole idea. Is one model better than the other as we go forward with implementation for CCHD?

DR. GREENE: It's possible that I misunderstood. Absolutely I think it's an issue for the committee. I'm still not getting how -- at least one State has said it's absolutely not a lab

issue. It's a State issue. It's a newborn screening program issue. But it's not the lab.

DR. BOYLE: Can I just follow up on that point?

CHAIRMAN BOCCHINI: Sure.

DR. BOYLE: I mean, this may be just totally -- I was thinking -- make this comment because I wondering whether or not the name of the committee needed to be -- subcommittee needed to be changed from "laboratory" to be something relative to testing or, you know, the screening test since trying to incorporate point of care within the context of that. And I guess this discussion makes me think that maybe if that's not a moot point, maybe you need to think about the committee more as the committee that's overseeing the screening testing issues, and that the follow-up in treatment. And I know we deliberated about our name yesterday a little, and would really pick up from that, you know, after the testing procedure.

CHAIRMAN BOCCHINI: It certainly makes sense to broaden the title of the Subcommittee on

Testing Standards and Procedures. And that could easily encompass all of what we've just talked about. And I don't know whether that would require the Secretary's -- so we'll take that under advisement to see how that -- whether to bring that forward that way.

But I think we all know we've broadened it from the laboratory to all testing. Denise?

DR. DOUGHERTY: If what we're trying to do is pick one project from among these, my preference would be project number one, which seems most closely tied to the laboratory and testing. And it seems like it would be -- there really is a need, and it would be an excellent idea.

CHAIRMAN BOCCHINI: Okay. Any additional questions or comments? I think that's a good approach. I think you're right. I think that that's a project that looks like it's coming to a head, should be potentially done maybe in September from what we heard -- January. So that would be something that would be good to evaluate and see if we could adopt those and endorse them, and see if we

can help promulgate them. So I think that is good.

So out of these three, is there consensus project number one looks like the best? Okay, good.

And then priority three, any questions or comments about priority three?

(No response.)

CHAIRMAN BOCCHINI: Everybody is in agreement that this -- I mean, I think we've spoken about this before, and I think this honed probably the best at this point that it has been. And I think that it is something that we should be doing.

DR. DOUGHERTY: One thing is whether it's the Laboratory Subcommittee that should be doing it.

DR. COPELAND: During the discussion yesterday, the idea was not that it would be the Lab Standards because the idea being that the Condition Review Panel or the subcontract with the Evidence Review Panel will develop the process, be informed by Lab follow-up and Education. But we can definitely contribute to it because it's not just any one aspect of the subcommittee.

DR. MATERN: Yeah. I think the committee

can contribute here because, again, we're not screening always for just conditions. We're looking at analyzing our suggested conditions. So you can just take one out and you shouldn't consider the laboratory aspect of how you actually do that.

CHAIRMAN BOCCHINI: So it is true the Laboratory would be one aspect of it. And, again, if the committee was looking at specific things, the subcommittee could come back to the full committee, or we could make a decision the committee needs to look at that particular problem or process and move forward. But I like the idea that someone is thinking about that.

DR. MATERN: Actually I think in the ACNG report from 2006, they already indicated that there's this flow chart and how to do it. I think the question is, who is going to come forward and say, I don't like this disease on the panel. Please tell us why the heck it has to be there, or, could it be removed, or upgraded, or whatever.

CHAIRMAN BOCCHINI: Okay. Additional questions?

(No response.)

CHAIRMAN BOCCHINI: All right. Well, thank you, Dieter, and thank the subcommittee for its work. I think that's a very good start. And we may come back to some of the issues related to that subcommittee as we go through the others and look at some overlap between priorities.

So, Don, Subcommittee on Education and Training is next.

DR. BAILEY: Good morning and thanks. So our committee must not have been efficient because we met until 5:15, and we were still going strong. So a lot of wide-ranging issues to discuss.

So I already reviewed for you the committee charge, so I'll just move into what we discussed, and then on to our final priorities.

So I wanted to make sure that people understood that because we have such a broad charge that we have a broad constituency in our membership, which both helps, but also challenges us to make decisions.

So we have really four groups of members.

We got five committee members that are actually members of the Secretary's Advisory Committee. We have six members who are official organizational representatives to the committee. Additionally, we have two representatives from federally-funded grantees. And then we have consultant members to fill in additional slots or needs for the committee.

So in our last meeting we have -- we have 20 slots for the committee, and we have 16 members. So we went through actually a great process that Sara and her colleagues initiated, which was a self-nomination process that was a call for that was issued through a number of different venues, and list serves, and so forth people to nominate for three slots. One was for someone to represent -- hospitals and birthing facilities, we really didn't have anyone who was really deeply rooted in the actual hospital setting and what was going on there when blood spots are collected.

The second one was to -- because Andrea and Jana moved to the Education and Follow-up Committee. I guess they were given a better offer.

(Laughter.)

DR. BAILEY: We needed another -- at least one more parent representative. And then we needed -- although we had representatives from different professional organizations, we didn't really have one person who was kind of professional training was their primary responsibility.

So I've highlighted in red the new committee members. And I was really both kind of amazed and impressed and overwhelmed by the quality and the enthusiasm of the applicants that we had for this position -- all these positions. And so it was a difficult choice, but it was great to be able to make those choices. And I think we brought in some new people and some great input.

So first, So Allen Hogge has stepped down as chair of the Committee on ACOG. So Nancy Rose has now replaced him, and Nancy will be a great addition. ACOG is an important player in this whole landscape, and so we're real pleased to have her on the committee.

Emily Drake joined us as a representative

of the Hospital Birthing Facility Emily is a professor in nursing at the University of Virginia. She's also a member of the board of the Association of Women's Health an Obstetrics and Neonatal Nurses. And so she brings in a different organizational representative. And we had set a goal also for a nursing representative. We still hope to expand that in future membership slots, but Emily will provide a great representation, both from the birthing facility and from nursing.

Jeremy Penn is a parent of a child with SCID, and only in the last couple of years, and obviously clearly rooted in the experiences of what happens through newborn screening, become very active in his State. And so we're real pleased to have Jeremy as a part of our subcommittee.

And then Joan Scott is with the National Coalition for Health Professional Education and Genetics. And so we're excited to have her as well.

So Emily and Joan were not able to join us yesterday, but they will -- because of the late appointments and some other commitments and things

that came up at the last minute. But we're looking forward to having them in future meetings.

So the goals for our meeting yesterday were to, first of all, to review ongoing activities and get some updates from organizations and projects. And, again, that's a challenge for us because we have so many different people to update, and we want to know what's going on. And so we're going to have to figure out in the future how to balance reporting versus decision making and action items.

We also heard a preliminary report on whether States collect data on newborn screening refusals and whether State policies affect refusal rates.

We discussed potential collaboration with the Condition Review Group, and Alex Kemper came and joined us to provide guidance for advocacy groups and other regarding the nomination review process.

We reviewed findings and initial recommendations from the recent newborn screening awareness campaign strategy summit meeting. And we

discussed the awareness activities planned in association with the 2013 50th anniversary of newborn screening.

So in terms of newborn screening awareness activities, I'll just very briefly touch on this right now because I've been asked to do a more thorough report on this this afternoon after lunch.

So if you recall from previous meetings, we had contracted with Porter Novelli to do a phase one media scan, and this was completed, and the report was presented at a prior Secretary's Advisory Committee.

Then recently HRSA, working with Porter Novelli, we had a planning committee, which I chaired, to convene a strategy session to discuss strategies to inform and educate the public about newborn screening. Really kind of three major questions to try to address in that strategy session, which should be the focus of a major campaign: who are our audiences and what are the messages? And I'll come back and convey the results of that this afternoon.

We also heard from Carla Cuthbert at CDC about an update on the 50th anniversary plans. I think everybody knows that next year is the 50th anniversary of the first year that some States began screening for PKU. And so it's a great opportunity to both celebrate that, but use it as a vehicle for public awareness.

And so the CDC and APHL are taking major responsibility for that. The media scan and the strategy session that we had in April are being used to help partially inform that planning process, and there are a wide range of really interesting activities currently in the planning stages. And I think a number of them have implications for our entire committee. And, again, I'll come back and share those with you this afternoon.

We had some updates from Genetic Alliance, and I'll just report on two major things. One is that the Genetic Alliance has been organizing something called the Consumer Taskforce. And so this is parents who meet on a regular basis, both virtually and literally, to provide guidance in a

variety of different arenas. But three major goals. One is to help promote newborn screening at the local level so that the awareness campaigns that we're talking about with our activities are more at the big picture level, at the national level. But really a lot of the activity is going to occur on the local level and in one one-on-one interactions with various providers.

And the Consumer Taskforce is looking at different ways to promote newborn screening at the local level, to really identify the on-the-ground problems that might compromise or limit the benefits of newborn screening, and then help inform continued development of the Baby's First Test website.

And, Natasha, I believe that there was -- you had a meeting of this taskforce in the last couple of days, and a number of people here today are members of that taskforce. Are they still here? Maybe you could raise your hands? So great. So we want to just thank you so much for your dedication and commitment.

(Applause.)

DR. BAILEY: A number of them came to the Education and Training Follow-up Committee meeting, and we invited them to make comments whenever they wanted to, and they did. So that was great input. And so we're looking forward to continuing to work with that group and hearing the great work that you're doing. Also this is all funding from HRSA.

Genetic Alliance has been organizing a small grant competition called Challenge Awards. They have $100,000 a year set aside for these awards. And so at the last meeting, I reported that they had received a fairly large number of applications. We're in the process of reviewing those. We couldn't announce them. So now those awards are announced. There are six new awards. They hadn't planned on funding that many, but they were able to reduce the funding on some of them, and probably did some behind the scenes negotiations, Natasha. But six new awards have been funded. I won't go through them, but the list of them is available on the Genetic Alliance website.

The primary focus of these awards is on

developing and evaluating the effects of a variety of different educational materials. And so some of them are looking at videos. There's one looking at social media, one looking at print materials, web-based applications. And so this is very much aligned with the work of the Education and Training Subcommittee, and so we'll be very much interested to hear reports of those projects going forward because we think those are the kinds of activities that will provide better evidence of the efficacy of a variety of different approaches.

I think I mentioned yesterday that Michelle Lewis and Aaron Goldenberg had asked if they could present some preliminary findings of a project that they were doing on newborn screening refusals. And so they conducted an e-mail survey of the 50 State labs in D.C. And one of the major questions was, do you track the number of individual parent refusals for newborn screening? This would be in contrast with a more simple algorithm of just comparing the number of screening samples received per year with the number of births.

And at least from this report of the survey, a fairly substantial number of States don't track refusals in this individualized way. And only 14 States tracked the reasons for refusals. And this is a very complicated area, and some States are now allowed actually to ask parents reasons for refusals.

But this does point, I think, the limited data that we have that could be helpful. So a better system to track and report refusals at the national level would provide useful service surveillance information, and would allow for monitoring of trends over time, and especially if this were -- there was a rapid reporting of these. And also studies of the reasons for parent refusals and how those vary across settings or time would be very informative.

We think this relates to the Education and Training Committee because it all has to do with public awareness. And if people are opting out of newborn screening, and if that happens to increase over the next few years, we would like to know

quickly whether that happens and the reasons for it. So we don't have a specific goal or objective for this yet, but we wanted to highlight this as an important gap in our knowledge.

So Alex came in and talked with us, and, as I mentioned yesterday, we're exploring ways to collaborate with the Condition Review Group. And so what are the problems that we're trying to solve? Well, one is to increase public transparency for what we do overall as a committee, and the rationale for the decisions that are made ideally to provide feedback to nominators regarding next steps, and to support future nominators in preparing successful application packages.

So we had a lot of brainstorming discussion about wouldn't it be nice if. So wouldn't it be nice if we had short, plain language summaries of the evidence reviews so that people would have a distilled version of what decisions were made and why? Wouldn't it be nice if we had a blueprint for future nominators, so if you're thinking about bringing your condition forward for

the nomination process, we have the nomination review form. But wouldn't it be nice if we had a more extensive blueprint and guidelines for how do you maximize success there? Wouldn't it be nice if we could provide maybe more public friendly information on the Advisory Committee's website? Wouldn't it be nice if we created a lessons learned case study for future nominators? And wouldn't it be nice if we had a point person to help nominators navigate the process?

So these are all things, kind of blue sky thinking about, gosh, wouldn't it be great if we had these things available? We think it would help our work, and it would help the work of advocacy groups and nominators coming forward. So obviously each of these has resource ramifications, and so we'll come back with some suggested priorities shortly.

We had a couple of other brief reports. Beth Tarini gave a brief update on continued implementation on the activities of the Genetics and Primary Care Initiative. So if you recall, this is a three-year project. It's a cooperative agreement

with American Academy of Pediatrics. It's now wrapping up year one, Beth.

And so the goal with this initiative is to increase primary care provider knowledge and skills in providing genetic-based services. And so this is not related just to newborn screening, and this relates to some of our discussion earlier about are we just focusing on newborn screening things. This is a much broader initiative on primary care provider care knowledge and skills and providing genetic-based services. And so they're just getting this up and running, and we'll look forward, Beth, to a much more complete report.

And I think as we get, just like Dieter was talking about Piero, maybe coming and giving a report at the end of this project. I think certainly at the end of this project, we'll want a whole report to the Advisory Committee on it.

And we have a variety of activities going on at ACOG. Nancy Rose is developing a manuscript to provide further guidance for implementing the ACOG recommendations regarding newborn screening

information, dissemination to parents. And so we're looking forward to seeing that.

And, as you know, NUSPEG has been working on this project for family history for prenatal providers. And because of time limitations, we deferred the discussion on that, and we'll be having -- this will possibly be a whole Advisory Committee presentation either in September or in January because we think this project is being wrapped up, and it's now time to share the results of this with the entire committee.

So I presented our three priorities yesterday, and we've just tweaked them some, and tried to be a little more specific. But for some of them, they're still planning things.

So priority one is to continue to enhance our ability to track, provide input on, and facilitate the integration of national initiatives and committee initiated activities.

So in terms of our goals for the next year, first goal would be to continue to work with the professional organizations, but to be more

specific in terms of identifying priorities for newborn screening awareness efforts. So what would be the number one thing we would like for obstetricians to do? What would be the number one thing we would like for pediatricians to do? What would be the number one thing that would be most helpful in hospital settings all around this goal of increasing public awareness, and how we could make it a more systemic effort?

And then secondly, we want to conduct a scan to determine major education and training needs that would extend into areas other than newborn screening. And so our goal is that within one year we will have identified at least one major education and training goal that, again, addresses something outside of the newborn screening arena.

And so we've had some preliminary discussions about different examples. Certainly one of them would be we could set a high level of goals trying to reduce the time between presenting problems and a genetic diagnosis. And so that's not a newborn screening issue. It's when kids go back

to the pediatrician and are having either developmental problems or there are medical issues. How do we reduce that time between those presenting the first time the problems come to bear and a correct diagnosis. That's a complicated, multifaceted task, but that would be a good one probably for the committee and subcommittee to take on.

The second priority would be to continue to promote newborn screening awareness among public and professionals. And so we think we'll be very actively involved with collaborating with Carla and with the CDC and APHL as they plan the 2013 newborn screening awareness campaign. Again, I'll talk about that more this afternoon.

But a fundamental question is how and in what ways should our committee -- subcommittee and the Secretary's Advisory Committee be involved in these various activities, whether it's a day in Washington or whether it's public announcements and so forth. And so it's kind of hard to specify exactly what we would be doing over the next year,

except we will be in almost contact with them and seeing what we can do to help support that.

Secondly, to develop an action plan with specific objectives regarding professional practices in newborn screening awareness. I alluded to this already in terms of talking with the professional organizations.

So the fundamental question here is what changes in professional practice would most likely to result in increased public awareness about newborn screening, and how can we make those happen?

And then, third, and this evolved from discussions we had in this whole committee yesterday and then in our subcommittee, is maybe to try to identify a potential partner to develop a plan to inform State legislators about our Advisory Committee and about the evidence review process. And we think this can tag into priority three, which is developing more lay-friendly summaries of the evidence reviews themselves. So this would be to partner maybe with the National Conference on State Legislators or some other group.

So then priority three would be to provide better guidance for advocacy groups and others regarding the nomination review process. So there'll be two major sets of activities. Alex and the Condition Review Group are already talking about developing public-friendly summaries of previously-conducted evidence reviews, and we think we can help with that. And I think a major thing that we could do is to look at them as they're developed and give them feedback on the readability and clarity of those documents and think about ways that they could be disseminated.

And then secondly, we have created a subcommittee to recommend strategies for supporting nominators and advocacy groups with a goal here of increasing the clarity of the nomination and review process. I think it's pretty clear to us what happens, but to make sure that it's clear to the people who are bringing things forward. To think about providing guidance for, again, getting your condition ready for the nomination and review. And then thinking about ways to provide feedback on next

steps and at least in two situations. So for nominating conditions that we say are really not ready to move to the evidence review process, what feedback could we give those nominators to help get it ready? And then for review conditions that actually go through the evidence review process, but we recommend are not being added to the required panel right now, good feedback on what would be things that would be needed to help support that decision?

So that concludes our report.

CHAIRMAN BOCCHINI: Thank you, Don. That was a very good, strong reported. Very thorough, and I think highlighted things very well about all of the different aspects that the subcommittee is involved in. So very, very helpful.

Questions or comments?

(No response.)

CHAIRMAN BOCCHINI: All right. Well, let's then go back and look at each of the priorities and see if we can hone down a bit. And I'm assuming that all of these goals cannot be

worked on at the same time. So let's sort of hone down a bit and see what we think as a group would be the highest priorities for each of the -- within each of the priority goals.

And I'm very impressed that we have very strong interactions with ACOG and AAP and AAFP with the representation that we have. And I think that it's very clear that a number of projects have developed very nicely, that our approaching pediatricians, and family practitioners, and obstetricians to sort of modify what the current approach is to genetic diagnosis and neonatal screening. And so I think we have very strong relationships. And I think we can rely on them to help us identify those priorities and help with each of their organizations promulgate the information and see if we can change culture. So I think that seems to be going quite nicely.

DR. BAILEY: Again, related to that and topics we've brought up before is a major nursing organization representative would be really a key addition to us. And so we'll need to think about

whether that comes through an appointment to this committee, or whether we should just move straight to an appointment with our committee. Emily provided some of that with her association that, but we think we need also a larger nursing.

CHAIRMAN BOCCHINI: And hopefully with this first round of applications for liaisons that we will potentially have a nursing partner for that.

DR. BAILEY: That would be great.

CHAIRMAN BOCCHINI: Okay. And then education and training needs into areas other than newborn screening, I think that, you know, we're all -- I think that was a very good thought, and I like the way you put that. And I think that's an easy thing to start working on. I think that would sort of help lead us towards the next stage of development of our overall committee, and I think -- to me, that's a very worthwhile project.

DR. BAILEY: You mean the goal that I said of reducing the time between --

CHAIRMAN BOCCHINI: Yeah, to find -- right. To pick a disorder or to look at reducing

the goal of reducing the time from onset of symptoms to diagnosis. And then using that as sort of a guide on how to go forward as a committee, and whether we can make that something that would be a public health issue, or would be something that could lead us towards diagnosis outside of the newborn period.

DR. BAILEY: So maybe take a prototype condition then, that would be a good example, rather than trying to do an entire -- solve all the conditions at one time.

CHAIRMAN BOCCHINI: Right. Yeah, Coleen?

DR. BOYLE: Just to go back to the first working with professional organizations, just something about the conversations yesterday.

CHAIRMAN BOCCHINI: I'm sorry, the --

DR. BOYLE: I'm sorry.

CHAIRMAN BOCCHINI: Oh, no, no, no, no, just your first bullet under goals for next year. So with professional organizations, you know, this is clearly a two-sided conversation. So, I mean, it's one thing to work with the professional

organizations, but somehow looping parents, you know, who are part of that conversation, just get their sense of the needs relative to those different stages where they would be interfacing with the professional. I think that's really an important part of it. I'm assuming that's part of it, but it's not explicitly --

DR. BAILEY: That's one of the great things about the committee. Now we do only have two formal parent slots on the committee, so hopefully in future meetings we'll continue to have an audience that can help us with this as well. And I think our linkage, we can have more formal linkage with the Consumer Taskforce through Natasha to help bring that perspective as well. But, you're right, the people who are being served by these professions need to give input on what would they like to have approved.

CHAIRMAN BOCCHINI: Beth?

DR. TARINI: So I wanted to echo Don's point that actually a lot of the conversation around parents came from the audience members, who were

parents who had experiences. So that was very helpful.

I don't know the extent to which -- trying to be a pragmatist -- we're going to be able to do that as well as the professional orgs from our committee. So I know there are many projects out there, Carol's and others, who are doing actual research with parents.

So the extent to which we can leverage those existing studies and that data might be more helpful than trying to get it ourselves. But definitely incorporating it in, but I don't know that it will be feasible or a good use of our time to spend getting to them, because I think they're hard to get to, and hard to get -- when you get to them, it's not clear that you're getting a representative sample.

DR. BAILEY: But it is certainly something that the Consumer Taskforce could help with I think. Natasha, would you like to speak to that at all?

MS. BONHOMME: Yeah. No, I think the Consumer Taskforce, as well as just our range of

other public groups that fall into that public category, we can definitely do that, though I do think it is something to consistently be thinking about.

I can't say that the whole Consumer Taskforce will be able to come to each meeting, but I do think that it is important to figure out what are interesting and new ways of bringing in that input, whether it is sending out questions beforehand so that we can at least make sure that there is some level of engagement, even if it isn't a whole group here physically every time. I mean, I think that would be ideal, but that isn't necessarily realistic or in the plans at this point.

But I do think being able to reach out frequently to parents, whether it is the Consumer Taskforce and to be on that, you know, who the members of the Consumer Taskforce represent, can definitely be done, and we should always be thinking about, like I said, creative ways of getting that input.

DR. BAILEY: And there are other parent

organizations that pretty frequently come to these meetings as well, and so hopefully we can rely on their input as well.

And I do agree with you as we kind of narrow down on specific things, we can be more targeted in the kinds of information that we would like from these groups.

CHAIRMAN BOCCHINI: All right. Let's go to priority two then. We had decided that the priority it look at the one major -- look at a genetic or metabolic disorder, the outside newborn screening area.

So I think clearly there's been a good interaction with the development of the awareness campaign in this committee, and now a good understanding of what the goals of the CDC and the public health laboratories are for recognizing the anniversary of initiation of newborn screening. And so I think that ought to be a priority that we should continue to work on because we need to try and be as much involved as possible and utilize this as an opportunity for everyone to raise awareness.

I think that ought to be a major goal for us. And the subcommittee is certainly poised to be involved with that.

DR. BAILEY: And it's got to be an immediate goal because it's happening.

CHAIRMAN BOCCHINI: Right. It's coming, and so I think that's a top priority.

And then the action plan, we've just completed the phase one of the newborn screening awareness, and I think this is important for us now to decide. The committee had decided to complete phase one, get the information, and Don will discuss that in more detail this afternoon. And I think we ought to consider then what sort of approaches we should have to get into phase two. But very importantly, consider what's going on with the 50th anniversary and use that as an opportunity to sort of include that into what we're thinking. So I'd like to sort of fold that into number one there, the first one.

DR. BAILEY: It certainly will be there, and there will be some integration. But I think the

primary goal of the campaign is not going to be targeting professional practice as much as it is other activities, but it will be a great springboard. And this second one is really, like, after the celebration going forward, although we don't delay discussing this. This really talks about institutional changes.

CHAIRMAN BOCCHINI: Right. But these are changes in professional practices that would improve public awareness, right?

DR. BAILEY: Correct.

CHAIRMAN BOCCHINI: So I think we need to be sure that that -- right, okay. Steve?

DR. MCDONOUGH: Yeah. I'd like to make a suggestion to the committee that our fall 2013 meeting would be held in conjunction with this event if it at all possible. That the Secretary of Health and Human Services, Assistant Secretary, Surgeon General, head of CDC, possible NIH be invited to the event. And that our committee meeting could be held in conjunction with the event.

That organizations such as March of Dimes

of Genetic Alliance, if that they have the ability to recognize legislative champions across the country or in this area, Washington, D.C., that there could be events that could be held at that time to recognize them.

There's a great potential for the whole history of newborn metabolic screening going back to 50 years to have a tremendous celebration here, and to capitalize on the opportunity for going forward to screen for new conditions, to deal with threats to the screening process.

So anyway, for those of us who have practices and busy schedules and stuff like, to be able to come out and participate in that rather having meetings two weeks apart or a month apart. We've got a year from now to set the date, and I think it would be -- there's so many players that have a history in this, and they're recognized. All the efforts of CDC, and March of Dimes, and all the other organizations have been at this for a very long time, in addition to HRSA.

Anyway I think there's just a lot of

opportunity. We got a year to do this, and just bring that up as a possibility to do a lot of coordination. And I think you could get some pretty important people there at this event.

DR. BAILEY: So what Steve's referring to is, which I haven't actually described yet, is a plan, kind of one day in Washington that CDC and APHL are considering as a part of their plan. And I'll report on that this afternoon, and maybe we can continue that discussion after we contextualize it in the context of that report.

CHAIRMAN BOCCHINI: Comment from --

MS. BAKER: Mei Baker from Wisconsin. Yesterday I heard committee members talking about when we promote newborn screening and we increased awareness, also, again, the balanced information and also recognize that it's challenging. And after that, I think the newborn screening, if we use this opportunity to try to use the lay language to educate the public and also primary care, to think about newborn screening really as a risk assessment is not a dynastic arena, but that was a diagnostic

arena.

Also people said, well, you can be diagnosed by newborn screening or diagnosis. I think this is needed because I think the expectations are different. The expectations are right that people you say, okay, the purpose is we want to make sure the majority group we put aside have the very, very low risk and have small groups. We want to take another look.

So this way, I think this opportunity kind of gets to this concept of the primary care and also the public.

CHAIRMAN BOCCHINI: Thank you. Jelili?

MR. OJODU: Good morning. Jelili with APHL. Thank you so much for that suggestion of having this meeting coagulated with the event that we're planning in 2013.

The Atlanta event, I think most of you know, that's going to be held on May 5th through the 10th in Atlanta. And I know the logistics of moving Federal meetings can be a hassle. But that is an option. I know the Secretary's Advisory Committee

is already scheduled for the 16th and 17th of 2013.

And then we're planning to have a Washington event that's specifically geared towards lawmakers, and advocacy groups, and parents to, as someone rightly noted, to highlight the achievements of newborn screening, and to bring a lot of attention to a number of things, including the fact that we need to reauthorize the Newborn Screening Saves Lives Act.

So we plan to have that in the fall of 2013. We haven't set the date yet, but normally it's tough to have a meeting like that before the end of the Federal Fiscal Year, which is September.

So we're looking at October, but we can certainly be flexible in making sure that we can have something together with you all. So thank you.

CHAIRMAN BOCCHINI: Thank you for that comment. I think that would be great if we coordinate things so that the committee would be in town and be available to participate in that in whatever ways possible. I think that would be great. Make a comment?

MR. MORRISON: Hi, yes. My name is Bill Morrison. I'm with Consumer Taskforce for Baby's First Test, and I'm a parent advocate. And I don't mean to get real basic on it and everything, but a lot of us advocates, we make tee shirts. We do things. Have you all given any thought to something as basic an official tee shirt for the 50-year anniversary, that kind of thing? You know, even maybe have everybody wear them to the celebration and have a great photo op? The kids would really get into it.

DR. BOCCHINI: I think that's certainly a reasonable suggestion, and I think advocacy groups, certainly I would believe, and I'm sure we can hear from the CDC, would be welcome to do things like that for each group to be represented and have whatever they decide to be representative of the group, to be involved in that in some way. So I see it wide open to do those kinds of things.

MR. MORRISON: Maybe a contest for some of the kids that have been tested and have been identified, and you could come up with a selection

for one official shirt for the whole Nation, that kind of thing.

DR. BAILEY: Sure. That's exciting. Thanks for that suggestion, yeah.

CHAIRMAN BOCCHINI: All right. Other comments?

DR. BAILEY: I'm sorry to keep saying we'll talk about that this afternoon, but we actually will.

(Laughter.)

CHAIRMAN BOCCHINI: We do have a more detailed presentation coming up. Okay.

DR. BAILEY: Do you want to talk about the State legislator question at all? I mean, we have to figure that out, I think.

CHAIRMAN BOCCHINI: Yeah. We're early with that, but I think it did come up in yesterday's conversation, and it would be important for us to be a reference site and a resource for individuals who are facing things in legislatures. And also to turn around and be able to inform legislators in a prospective way to identify issues for them. And I

think that might keep things in better balance. So I think that should be maybe more of a longer-term goal that we should have.

And then I think these are really good ideas, and I think would enhance the work of the committee overall. And I think these are -- I think it would be -- I don't know how recently we've looked at the website and are comfortable with the way it is, but I think having a formal review and recommendations to make the website more user friendly, and seeing whether we can enhance the guidance available for people who wish to bring forward a condition nomination, and then making sure that we provide them with good feedback as to what was missing.

Now we currently send a letter, which details the discussion and what the committee feels is missing. But whether that's enough or not would be good to go back to some people.

DR. BAILEY: We think going back to some of the nominators and asking them about their experience and what would've been helpful and so

forth would be a good idea.

CHAIRMAN BOCCHINI: Yes. I think that's a good project, and so put that as a middle sort of project. I think having that done at a reasonable time frame, maybe within the next year would be pretty reasonable.

DR. BAILEY: I think that's all possible.

CHAIRMAN BOCCHINI: Okay. All right. Other thoughts related to that?

(No response.)

CHAIRMAN BOCCHINI: Okay. And then that should do it.

DR. BAILEY: That's it.

CHAIRMAN BOCCHINI: Thank you very much, Don. Appreciate it.

Okay. Carol, we now have the Subcommittee on Follow-up and Treatment. And Carol Greene will give the report.

DR. GREENE: Okay. Kind of heading into the lion's den here. We met until closer to 5:30, and we were still going strong. And we really appreciated that right around 4:30, Sara wandered in

with a message for us, and I took advantage of it and had her remind us that we had to end up with something focused for this committee. So I appreciate it greatly.

And I want to also say publicly that I felt like I had to keep on cutting on very rich discussion to keep focusing us so we'd be ready for this discussion here today. Clearly we have passion and ideas, which are some of the really important resources that we need to address important questions, and to begin to look at -- to be able to go forward with some of the projects that we're going to be proposing here today.

So I finished my slides just a short while ago and did not try to prepare slides that summarized the very rich presentations we had.

So I want to start by saying we did review our priorities and projects, and that's what my slides will focus on. We worked first on understanding and focusing our priorities, but I'm going to switch all the way to the very last slide here just so that you can see the committee members.

And we introduced ourselves. We welcomed new members and embarrassed Coleen with some really cool information and thanks that came from various people.

I should probably mention that is from official because have to make a request, but by means of saying this, I am making a formal request to have Chris Kus as co-chair of the subcommittee. But he did agree.

So let me leave that up just for a moment while I say that we heard -- first we heard updates on two projects. One is not our project, but a project done by another program. And we got a chance to look at a manuscript on Medical Home that is going to be presented here, and we want to say publicly that we very much appreciate the opportunity to hear about the work, and that it includes some ideas that resonate with our proposed projects that we can look at to improve communication and care coordination to see how things are going and where there might be opportunity to work.

We also heard update on the manuscript that was developed from work of the -- proposed initially by subcommittee led by Sue Berry and a number of other people who worked very hard on a manuscript now on medical foods. And you'll be hearing about that today as well in the full committee.

We heard from -- and I want to say that we gave people very, very, very short notice to put together their presentation, and they were wonderful and appreciated. We heard from Andrea Williams and Alexis Thompson. We've been exploring whether and, more specifically, why and how sickle cell would be an important case to focus projects on that will help inform broader questions, important in and of itself, but also inform for other disorders.

And then we heard from Coleen an update -- "update" I think is probably the wrong word. A review of how far we had gotten in exploration of roles and responsibilities in follow-up and treatment. And we explored how that prior work could help inform future projects. And then we had

a brief overview from Chris about public health involvement in long-term follow-up and treatment.

So we worked on understanding -- these are fundamentally -- these are the same priorities that you heard yesterday from Coleen, slightly reworded to avoid some of the lack of clarity that we turned out to have, so that using the words "impacts" and "outcomes" and looking -- basically it turns out we had some different understandings of what we meant by those priorities, and we did a little bit of wordsmithing. And I hope that the people who were actually there will let me know if I've failed to capture what I think we were looking for.

Then our first priority slightly restated is to look at -- and we had originally the word "facilitating," but we're really not facilitating. What we're trying to do is understand, to learn what's been going on, and to try to do it in a timely fashion so that -- and this is a recurring theme so that what we learn from the past can inform the future.

Our second priority -- I think it used to

be closing gaps in access, but access is a part of the broader system, so we reworded it so that in that second priority we're really focusing on understanding the process. And it was pointed out repeatedly that having a good process doesn't tell you for sure that you have a good outcome. And so our third priority is really to focus on are we doing good things.

And in a sense, we agreed that's really the focus of the whole committee, but we really wanted to make sure that we're capturing something important.

There are a couple of things that we also had promised to discuss during our meeting that got sort of kind of table, but tabled with nuances to it. So we realized it would be difficult to change the name. We discussed the proposed name change. There was a sense that we liked the idea of emphasizing quality, but that we can do that in our priorities, and many people said we worked really hard to make sure that we have "follow-up" and "treatment" in the name, and we wouldn't want to

lose that. So we're perfectly content to not ask for a name change, and just to make sure that we're very clear in the priorities, what we want to focus on.

We did ask ourselves if we want to go beyond newborn screening. I will say on a personal note I was very glad to hear the Education Committee say that somebody's going to go beyond newborn screening. But as a committee, we're not ready to do that yet because it was very clearly stated that we have things that we want to work on first, and that can then inform the future. And we did agree we'll revisit it, but not now.

So an overview of what we discussed. We're really -- and these are recurring themes in what we're going to come back to in terms of our priorities and proposed projects to address those priorities. We're really looking in different ways at learning from the past to inform how people will go forward. And we're focused over and over and realizing what the subcommittee can do, that we're not in a position to simply take on large projects,

but what we can really help to do is frame what are the questions of interest, and identify sources of data, and identify where there are gaps so that we could help to point towards opportunities to coordinate across existing projects and programs, for example, in long-term follow-up, and asking what are the right questions, link with the NBSTRN that are working on framing questions in looking at some specific projects.

Again, we're going to come back to the idea of sickle cell as a test case, looking at ongoing HRSA projects -- and forgive me if I get some of the names wrong, but RUSP projects and the Secretary's initiative, and to build on what's available, and explore what is still needed.

We're trying to create suggestions for projects that the project in and of itself will intrinsic value for that condition or for that piece of the process, but also that we want to design projects that will help us to model not just so that what we learn from one condition can assist us with doing a better job with other conditions, or what we

learn from one part of the process for one condition can help with other conditions as well.

We want to add value to ongoing projects, but we also want to take advantage of the newly-developing programs and where there might be an opportunity to start things right with what we've learned from other programs.

And our next steps obviously after input from the full committee, we have some workgroups that are poised to flesh out some of the proposed projects.

So the next three slides, each one has the proposed projects within each of the priorities. And I should probably also say that as I was preparing the slides, I realized that we, I think, have done a very good job identifying some projects, which hopefully will be of appeal to the full committee that will allow us to use broad projects to inform different goals. So some of our proposed projects are different cuts of a larger project.

So in the goal of looking at screening program implementation, we're looking both at short-

and long-term follow-up and treatment. Questions that are important are, where we can help to inform the questions, what are or what should be the metrics? An example of a metric that was mentioned in the context of CCHD, for example, would be should children who have CCHD have developmental -- formal developmental evaluation at age two. And specific metrics need to be focused on the process as well as the outcomes, exploring what are the costs, what are the impact on families, and the idea to explore current and possible models.

There are three possible projects up there. Those are fundamentally the same three projects that you saw yesterday morning. "Hearing" is in bold because there were -- I can't say there was actually consensus. There was a lot of, to some extent, consensus that hearing screen is important to look at because it provides a model for point of care. It provides an opportunity to look at some of the different elements of that system. That's one where I said earlier I think it overlaps with one of the proposals from the Testing Standards Committee.

There were a lot of votes for looking at -- not votes. We didn't take a vote, but a lot of support for the idea of looking at CCHD because it's being implemented now. And there's opportunity to make some -- to have some impact there. So it would be perhaps more straightforward to do a project. Again, we have people poised to clarify -- to design what we would look at. But hearing screening got a lot of support as being a test case, and having said that, there is a, I think, a goal that we would be able to use that information to feed back into the development or the current ongoing implementation of CCHD. But we hadn't gotten into discussion of that.

Our second priority, closing gaps in systems of care, possible case studies and projects. There really are two possible projects here, and then something that we would like to hear about at -- we hope at the September meeting. So let me take that one first because that's not -- the bottom bullet is not actually a proposed project. There was some discussion and some recognition of the fact that depending on what the Supreme Court decides,

there could be profound changes in the way that care is delivered to children.

And we have been hearing certainly Maryland and New York, both Chris and I have been hearing to expect, the word was "profound" or "extreme" cuts in the safety net programs because if the design goes through, the idea is that all care will be delivered in the community locally, and we won't actually need safety nets because all the vaccinations will be happening near home. And if they're cutting all the safety net programs, that cuts some of the specialty. So we have been told at the State level to expect profound cuts.

And we think that it would be a good thing to hear about this in the full committee. And, again, this not a project, but it's a request to the full committee to put on the agenda to hear from some people about what kinds of things we should expect to have to deal with as we're helping take care of kids with heritable diseases.

But back to projects. We've already discussed briefly that we'd like to do a case study

probably of hearing screening, but it could be of congenital heart disease, depending on what we hear from the full committee. You're going to hear in the next slide about our proposed case study of sickle cell, which is our most fleshed out proposal.

And we would like to explore as part of those two cases, explicitly explore what are the current and what is the variability, so not necessarily doing a State-by-State survey, but, again, we have a workgroup that's poised to design how we would actually flesh out the proposal. But as part of the two case studies that we want to look at, one for implementation and one for outcomes, we also want to look at the current -- how roles and responsibilities are currently played out, and what is the variability, and how can we use that to understand, for example, what might be points to consider.

We're not expecting to take up exactly where we left off and try to say what the roles and responsibilities should be, but to explore how those discussions can take place.

And we also thought about the possibility of a very focused case study looking at the electronic medical record, and newborn screening results in the EMR, and the EMR as a source of long-term follow-up data. That's another possibility, but, again, you see in bold the one that we thought was at the level of subcommittee discussion, the one we thought was important and would fit into the other proposals that we have.

And then my last slide -- except, of course, for the membership of the committee that we already looked at -- is our last priority, real world impacts and outcomes. And the second bullet is really looking forward to the future. It's the idea of making sure that we are poised to either look at other disorders, or that what we do with our proposed case study would inform how other people could look at other disorders. So the second bullet is really for the future.

We really have one proposal for right now of a case study. We spent some time thinking about -- and we are absolutely convinced that sickle cell

offers the opportunity to explore. It's one of the conditions for which we have been screening for -- you know, it's not the longest period of time, but it's up there.

We have lots of data across the life span. We have lots of sense of obstacles. We have an opportunity to look at whether we can document the improvement in clinical outcomes that was promised by newborn screening. It's an opportunity to look at carriers and variations. There's rich data sources. There's a lot going on.

There's opportunity for this committee's work to help coordinate work of other entities. And our focus -- what we would bring to this that would be special and helpful would be an opportunity to focus on developing key questions, understanding the data sources, identifying gaps in data sources. This, again, would -- thinking about what data -- what questions should a State be able to answer and can they? But really coming back to the outcomes and is the data there?

CHAIRMAN BOCCHINI: Very good. It's very

clear that your committee worked very hard to put this together. We really appreciate that. Thank you. And a very nice presentation. It makes very clear what the goals are and some real potential things that we can do to move ahead.

And I think this focuses on two things that I think were very important. One is we need to know what happens when we make a recommendation, so implementation is really important. And to follow up on that I think should be very important.

DR. COPELAND: We actually have an independent study ongoing on the impact of all the recommendations made by the committee. So we have the resources, and we're already going to start doing that. So you can cross that off your list. Good idea. Great minds think alike.

DR. GREENE: Which one?

DR. COPELAND: On the review of the impact of all recommendations.

DR. GREENE: Oh. I don't think that was one of ours.

DR. COPELAND: Okay.

DR. GREENE: I think I understood Joe to be saying that this might help flesh out an understanding of that.

DR. COPELAND: Oh, I'm sorry. I was premature.

CHAIRMAN BOCCHINI: So the other thing I was going to say is that I think this also speaks to partnerships because I think that some of the things in terms of implementation -- and I see not only -- I think the committee had made two recommendations. The most recent is critical congenital heart disease, but right before that it was SCID.

And so I think that you have a new laboratory procedure and a new set of follow-up requirements. And then now a point of care test with its own follow-up requirements.

And so I think we have two opportunities, again, potentially with partners -- public health laboratories, maybe the CDC -- to look at what's happened with implementation, and what the barriers have been. And both really speak to different aspects of what public health laboratories might

require and what the public health system requires to make them work.

So I think this would be really important for the committee, but I think we need some partners to help us do that job. And so perhaps just going back a little bit further, how do we partner, and who should we partner with, and who can we partner with to sort of get some real-time feedback on what's happening with the recent recommendations that we've made in terms of implementation, and then what does that mean for follow-up I think is really important.

So I think going back to -- well, let's get some other input.

DR. GREENE: If I could just say that's very helpful, and some of the rich discussion, which I didn't have time to explore, really did focus on exactly that question. Who do we partner with? How do we partner? And our role we would see as how do we frame what are the questions that need to be answered, and that will help us decide who we partner with.

And I forgot. My note said that I was supposed to say that recognizing SCID is important, but it is clear from our top two heart disease and hearing screening follow-up that the subcommittee was definitely interested in looking at implementation for point of care testing. That that was something that -- I think the sense was those were our top two that we were interested in looking at because of some of the special issues of point of care.

So recognizing the importance of SCID, but heart and hearing, hearing as the longer in existence point of care, that we were interested in looking at that.

CHAIRMAN BOCCHINI: Good. Questions, comments?

(No response.)

CHAIRMAN BOCCHINI: Okay. Then I think out of these three, and then the consideration of -- looking at implementation of SCID as well, again I think determining who to partner with and how to get the data so that we can have some feedback through

the subcommittee to the full committee, or directly to the full committee based on discussions that the subcommittee had in development of a protocol or program to do that, I think would be very helpful to this committee, and would move us ahead.

And I think Sara's comment is important, too. We need to know what is already going on so that we could perhaps benefit from learning from other agencies, and get information that's already potentially available to us if we knew who was doing what. So I think that would be important.

Oh, and then the second thing that I think impressed me was that I think some of these questions certainly can potentially drive some research. And so the question is, who do we partner with, and I'd like to hear from the Federal agencies that are around the table to help inform potential RFAs or other potential grant or contract proposals to consider looking at some of these questions. Are these things that we could work together to sort of prioritize as potential things that need to be addressed? Maybe we can have some comments on how

that interaction could occur.

DR. BOYLE: Well, I'll let Denise go first. I'm only teasing. Just from my point of view, if the work of the subcommittee could come up with some important questions, then, yeah, we definitely could consider that. Obviously we have an interest in hearing -- at least my center an interest in hearing screening, which is what I can speak to. And congenital heart defects I would have to talk with Chris Portier and Carla in terms of SCID implementation. But definitely that would be of interest to have that guidance.

CHAIRMAN BOCCHINI: So you are a potential partner to work on hearing screening follow-up. That's great. Melissa?

DR. PARISI: With regard to NIH, as I might've mentioned yesterday, the Newborn Screening Translational Research Network, which is a contract from NICHD to the American College of Medical Genetics, is actually supporting development of a long-term follow-up tool that I think could be accessed by investigators who may be wanting to

pursue studies looking at long-term follow-up for a number of these disorders.

So the goal is not reinvent the wheel, but to utilize resources that already exist, and that is one resource that would be available to the investigator community as well as many of our other Federal partners.

CHAIRMAN BOCCHINI: Great. So that's another --

DR. DOUGHERTY: Well, I'm not necessarily offering because we have a program called the Action Too Network. So one of the purposes of that is to investigate if you tried to change something in a system, to see what the effects might be. So it's kind of a rapid cycle evaluation network. It probably wouldn't meet the standards of research, but it's a contract mechanism, and it has, I don't know, 10 or 12 main contractors. And then some of the contractors have lots of hospitals that serve children associated with them. So that might be an opportunity. Not that ARC has the money to support a study like that, but if something came up, maybe a

bunch of us could cobble something together.

CHAIRMAN BOCCHINI: Okay, thank you. All right. Microphone?

MS. CARUTHERS: Hi. My name is Ruth Caruthers. I'm from West Virginia, and my son was born with heart defects in February of 2011. And where you were saying you wanted to know who to talk to, I helped get a law passed in my State that requires every baby to be tested for heart defects. It's named after my son.

And we worked very, very closely with the American Heart Association. I know the March of Dimes is helping in a lot of other States. New Hampshire's bill just passed through the House yesterday. We're having a lot of action. New Jersey, Indiana, Tennessee, Virginia, West Virginia obviously, Connecticut.

It's moving quite fast, and I'm really excited about that. But I just wanted to bring that up.

CHAIRMAN BOCCHINI: Thank you for that comment. And I think that there are certainly for

advocacy groups, they certainly are involved and may have a good pulse on a number of the things that are happening. So that's good. That may be helpful to us as well. Carol?

DR. GREENE: Chris had to leave, but that reminded me, he would certainly say that ASTHO and AMCHIP are going to have some important perspectives on, as you mentioned, States -- thank you -- that they'll have some perspective.

CHAIRMAN BOCCHINI: So I think there are a number of resources we can tap into and potentially get the information that we need, and then potentially look at ways to encourage research in particular areas. That would be helpful. Okay.

DR. GREENE: And I think that what we just discussed applies to projects within all of our three priority areas. You know, there might be different specific projects, but those are still partners that are important. And to that list of partners, I would say we probably need to add the hospital associations and the insurers.

CHAIRMAN BOCCHINI: Okay. Can we go to

the next slide? Yes, I think that with Coleen's comment, I think perhaps we can get together and discuss potential things that are already available, and then what might need to be put together so that we can sort of get a good feel for what's going on, and what the short- and potentially long-term follow-up.

DR. BOYLE: And obviously this is a shared program with HRSA, so there's a lot, I know, that we talked about yesterday about what's going on from the shared program responsibilities.

CHAIRMAN BOCCHINI: So I think that would be a really good start. Oh, then the last point about the electronic -- the HIT. I think that, like, we said, we're in the early stage, and that might be something that would be a later project, but one that could potentially benefit by looking at one of the partners and seeing if there's availability of research funds or something that would promote that being done in particular areas of pilot study of some sort.

DR. GREENE: So not to move forward with

such a project now, but to keep it on the radar screen and work on how we might design it and who we would partner with.

CHAIRMAN BOCCHINI: Right. And going forward with the first two as being primary projects for the moment.

DR. GREENE: I'm sorry. You said our first two?

CHAIRMAN BOCCHINI: The critical congenital heart disease and screening follow-up.

DR. GREENE: Okay. I just want to be really clear. So would the sense be that we would work on hearing screening follow-up, and then looking at the program implementation? And I'm not sure that we have an idea of a time frame. We'd have to work on it, but that once we have that project, we could turn it around and apply it to heart disease?

CHAIRMAN BOCCHINI: That sounds good.

DR. GREENE: Great.

CHAIRMAN BOCCHINI: Okay. And then here case studies -- I think we probably need to get a

better understanding of what is available and what's going for projects related to children and adults with sickle cell disease, so that you could determine how to then proceed. So maybe it would be sort of, again, to get an idea from HRSA and others what's currently happening, and see if that provides you with the kind of information that are considering looking for in terms of adequacy of follow-up and other aspects that you are identifying as important things towards long-term follow-up.

DR. GREENE: That would be an important starting place. I think our focus here is to -- it would involve other partners as well, and especially in this case it would involve, I think, ASTHO, AMCHIP, parents.

Our goal here is to understand how things are currently being apportioned, who's responsible for what, including who pays for what. We don't feel that we can undertake to say any "shoulds" here, but we'd like to understand the landscape, what is happening and where are there gaps.

And so this would -- not look at shoulds

at this point. It's looking at what is and looking at least two, starting with the hearing and the sickle cell because I think we know we're going to have consequences.

CHAIRMAN BOCCHINI: Right. I think, yeah, the continuation of the hearing is probably the same. Everybody in agreement with that approach?

DR. BOYLE: I just would add one thing, that's the second bullet or whatever, second indentation under roles and responsibility that's not highlighted. I see that as embedding these metrics, and somehow maybe roles and responsibilities in the context of electronic medical records is a way to make it happen. So I think, to me, that's almost the objective in many ways. So we need to keep that as our end post there because we want to make a difference, and I see that as one way of making a difference without trying to do it, you know, one condition at a time.

DR. GREENE: So to make sure that we include some look at EMR and the -- that actually ties in with what we had on the first slide is to

keep EMR very much on the radar screen as part of this.

CHAIRMAN BOCCHINI: That's a good point. Okay. All right. And then for here, I think sickle cell is a test case. Applying what we just said, you are also -- I mean, this would be part of what you would be doing for the sickle cell part. So that I think this fits in with what we did before. So it's the same project.

DR. GREENE: That's why we said that there are overlapping projects, and we're looking at addressing different important aspects, but we can use some concerted work on sickle cell and on hearing screen with keeping EMR involved in our thoughts, work with the same partners, gather the data, find the gaps, and answer three different kinds of important questions is our goal.

And nearly a third -- actually either nearly or more than a third of the subcommittee volunteered to work on the sickle cell project.

CHAIRMAN BOCCHINI: Okay. So I think that that sort of fleshes out your priorities. I think

that would be the goal. And then any other questions or comments?

(No response.)

CHAIRMAN BOCCHINI: Okay. So since Chris isn't here and he didn't have a chance to respond in a negative way, he is now your co-chair, okay?

(Laughter.)

CHAIRMAN BOCCHINI: All right. Any other questions or comments?

(No response.)

CHAIRMAN BOCCHINI: Okay, good. So we're about --

DR. BOYLE: Can I just ask a question?

CHAIRMAN BOCCHINI: Oh, yeah, sure.

DR. BOYLE: I think we brought it up when we started the Laboratory Standards reporting with the overlap between committees and sort of how we were going to try to get them to work more in unison. So I don't know if we'd want to have a little bit of a discussion about that, or if that's a later discussion point.

CHAIRMAN BOCCHINI: Yeah. Let's do that.

I think based on what we've done, we've eliminated a significant amount of the overlap that was potentially there from the prior discussion. So let's see if there's any other questions related, or let's discuss that further.

So I think we cleaned it up a bit, so I don't think we have a lot of overlap. And I think we have seen some opportunities for interaction from the committee that will be quite useful. But I think we pretty much cleared up the overlap. Carol?

DR. GREENE: So I would agree that you did because when Dieter was going through the prioritization of the proposed projects for the Testing Standards Subcommittee, the one that really overlapped with our implementation is not one that they'll be acting on, but they clearly had some good ideas. And if somebody from that group who was interested in that wants to work with us, I think that would be very helpful.

CHAIRMAN BOCCHINI: Yeah, and I think the fact that we've now sort of focused more on specific projects, that when the subcommittees then meet and

learn more about where they are with the projects and then present it to the entire committee, then other members of the committee who are on the other subcommittees can certainly weigh in and provide that feedback so that we can make sure that there is less of a likelihood of overlap.

So I think by doing this process, we've now focused the groups, identified areas of overlap that we have, I think, pretty much eliminated, and then provided the infrastructure here so that when these are presented and are moving forward, then the other subcommittees, through their leadership, and members, and liaisons, can help inform whether there is additional input or changes that need to be made.

DR. BOYLE: I was just trying to keep us all in our lane. I guess I was thinking more about synergies, that's all. Getting us to work more together to push things forward, and are there opportunities for that.

CHAIRMAN BOCCHINI: And I think that's a good point, too. Don?

DR. BAILEY: I don't have specific

suggestions right now, but I think we've done a nice job of eliminating redundancies, and kind of clarifying things, and showing where there's some overlaps.

But I do think there are still some cross-cutting issues that we've ignored that aren't a part of anybody's group, and I would just mention two of them, and I think I mentioned yesterday one is the ethical issues. And really no one's taking ownership of that, and there are some points I think at which we might want to have a broad committee discussion of that.

And the other is families. And so families are touched by each of our committees in very different ways. We are the Secretary' Committee on Heritable Disorders, and so not only are families dealing with having a child with a condition, but they're dealing with the heritable nature of these conditions. And we've not really had any discussions that I know of about -- and you mentioned this, carrier status, about genetic counseling, other kinds of implications for

families.

And so I don't know really where that fits in our committee and what would be formal activities that we would take. But I think those are examples of two things that would be some cross-cutting issues that we might in a future want to spend some time at least thinking about.

CHAIRMAN BOCCHINI: Yeah, I think those are good points. And what I had sort of listed as things that are potential topics for the entire committee that I think Sara and I need to sit down on and think through would be the issues that came up yesterday about what prior studies are necessary for us to consider as a group that something can go on to evidence review. I thought ethics issue as a broader issue that involves the entire committee, and that's something we need to think about as a committee.

I think point of care testing is something -- if we change from Subcommittee on Laboratory to Treatment Standards, it's still important, but I think that's also important overall for the

committee. And so I pulled that out. And then I think what you just mentioned is another one that certainly warrants full discussion by the committee.

So I think those I've sort of pulled out as being sort of overarching things that we may need to address overall as a committee. Carol?

DR. GREENE: And, again, in the very rich discussion we had, I think it's probably fair to say that there are a number of places -- I think many of the ethical issues, certainly not the blood spot storage, but many, many, many of the other ethical issues and family issues fit very importantly and prominently in what the Follow-up and Treatment Committee wants to do. That's why I wanted to make sure to include what's the impact on families in looking at the implementation.

And I would also say that roles and responsibilities is -- and one of the reasons that the prior work was so difficult, it's fundamentally -- it's talking about systems of care, but it's also talking about justice and fairness.

And then in terms of outcomes, we'll

really be talking about impact on families and, among other things, what are the gaps. And if you want to add anything either.

I think that families and ethical issues is an extremely important -- it's not even a subtext. It's kind of what we're about. I mean, it's about the science, and it's about the systems of care. But it's really about outcomes, which is about families.

DR. BAILEY: Well, this opens a whole can of worms. But I do think we ought to revisit this at some time. So when we did the ACMG scoring system, Mike, a number of years ago, we did add family benefits to family and society is one of the criteria for considering. And I know that generated quite a heated discussion in the field as to whether that was an appropriate thing to consider.

And I don't know how -- and certainly in our evidence review group -- evidence review process now, I think we allude to it, but it's never been a central feature of making a decision. And I'm not necessarily arguing that we completely review that

process. But I do think that -- I mean, clearly from the testimonials that we hear at every one of our meetings, families are impacted by these conditions, and screening does help families in ways that we don't often always consider.

And so I would just like for us to keep that on the table as an ongoing part of our discussion, as well as the sometimes challenges that screening gives when it discloses carrier status or other things that are surprises to families, or conditions that that we don't really know what to do with.

So there are benefits to families, and there are challenges that screening gives to families that I think we need to continue to have that --elevate that as a part of our discussion.

CHAIRMAN BOCCHINI: Yeah, we definitely do. And I think when Alex talks this afternoon about the current condition review process, I know in the past he's talked about when he's attempted to look at harms, there's really not much data there. But I think that is something that he has focused on

and something that we can talk about a little bit more this afternoon. But I think that is a significant part. Yeah. Coleen?

DR. BOYLE: This kind of continues on this same theme, and it's an issue I don't think any subcommittee can take on. It needs to be done at this level, which is, again, an ethical issue relative to what we heard yesterday with MPS I, with Krabbe. And there's some very serious interventions that are associated with these conditions, and very, very challenging decisions that families have to make with not a lot of information. So we're really trying to have a really better informed discussion about that.

CHAIRMAN BOCCHINI: Beth?

DR. TARINI: On the heels of Don's comment, as the committee moves forward to consider expanding their assessment, if they decide to move forward on considering expanding their assessment of benefits and harms, I would urge -- I'm speaking as an individual now, not as a representative of the AAP. I would urge them to consider the policy

implications of such assessments if they are to be folded into the decisions the committee makes because we know that newborn screening is a mandatory public health program.

And so expanding the benefits may have implications from a policy perspective as to how the program is implemented at the State level.

CHAIRMAN BOCCHINI: Thank you. All right. Other comments?

(No response.)

CHAIRMAN BOCCHINI: Okay. Well, thank you all. I think that this is a process that we started. Do you have a comment, Steve?

DR. MCDONOUGH: Are we going to vote?

CHAIRMAN BOCCHINI: Well, I think we originally talked about a vote, but I think we have consensus. But if we want to vote, we can. I don't really think that we need to.

DR. MCDONOUGH: It said on the agenda we were going to vote.

CHAIRMAN BOCCHINI: I know, and actually Sara discussed that before we started this process.

It did say vote, but I felt that if we came to consensus that we could all say if it's consensus, it's consensus, and then that's sort of a vote. All right. We're all in agreement?

(A chorus of ayes.)

CHAIRMAN BOCCHINI: Okay. All right. Then I think we have a 15-minute break now, so if everyone will be back here promptly at 11:00, we'll get restarted. Thank you all very much. This has been a good --

(Break.)

CHAIRMAN BOCCHINI: All right. Thank you all very much. We're going to now have a presentation by Alex Kemper. Alex is the Program on Health Services Research at Duke University, and actually is the leader of the Condition Review Workgroup. But now he's here to talk about manuscript -- medical home manuscript from Follow-up and Treatment Subcommittee. So he's going to present information related to this, and there'll be requests for the committee to vote, to provide its support for this manuscript. Alex?

DR. KEMPER: Thank you very much, Dr. Bocchini. And I kind of wish my department chair were here to hear that introduction because I got a big raise. I'm just the head of pediatric health services research, and I try to stay away from the people who provide adult care.

Thank you very much for allowing me to make this presentation on the medical manuscript that arose from the Subcommittee on Follow-up and Treatment.

What we would like for the Advisory Committee is to acknowledge the report, the title of which is *Family-Centered Coordinated Co-Management for Individuals with Heritable Conditions*. And specifically we request that the Advisory Committee review and acknowledge the enhanced description of the medical home and strategies for improving linkage to the Medical Home for Children with Heritable Disorders. And you'll probably all be happy to know that I'm not asking for any formal actions beyond that of you all or of the Secretary.

I'd also like to acknowledge the many

people that worked on this manuscript. Dr. Carl Cooley actually really led the product, but unfortunately he couldn't be here because he's in Hawaii, and I guess Hawaii won out over this.

(Laughter.)

DR. KEMPER: But we really had a lot of people who are both active clinicians as well as experts in genetic medicine. Dr. Ostrander presented this yesterday in the subcommittee, and I'd like to single him out for his excellent contribution and, you know, sort of keeping things real as an otherwise busy family medicine physician.

So I'm just going to describe the paper from the 30,000 foot level here just so that you have a sense of what's in there.

As you all know, in the 1990s, the medical home model emerged as a way of ensuring care for children with special healthcare needs. Of course the medical home model goes way back further than that, but in the 1990s this more enhanced definition arose related to the fact that medical homes should be comprehensive, coordinated, and family-centered.

There was an increased recognition that this approach may be beneficial for all individuals.

There are many, many definitions of the medical home, which I'm not going to go through. But I would like to highlight some of the activities of the various parties that are involved with the medical, and just touch on how this may be different for individuals born with heritable conditions.

So certainly in the primary care world, we would expect active coordination for these individuals with co-management. And as Carl defines co-management, it's really vertically within the healthcare system, so between the generalists, the specialists, and all the other people that are involved with taking care of these individuals, as well as horizontally across community-based organizations and resources that can help with individuals and their families.

But there are obviously other key stakeholders, including family care, again, getting back to primary care, which in -- in most cases, the primary care setting will be the site of the medical

home, so to speak. But as we describe in the paper, in rare circumstances, a specialty clinic or a specialty program may provide the medical home for a defined population and for some period of time. But in order to really do this, the specialty clinic or program would have to be comprehensive, longitudinal, coordinated, and accept first contact responsibility for all the things involved in care -- preventive care, acute care, and chronic care. But in addition, there's also the specialty care team that gets involved with all these different stakeholders.

In the manuscript care coordination is well described as being patient- and family-centered, assessment driven, team-based activities designed to meet the needs of children and youth, while enhancing the caregiving capabilities of families. And the care coordination addresses the interrelated medical/social development behavior, educational, and financial needs. So obviously it's not just a little teeny thing. Lots of work to be done.

One of the unique things in the paper's description is the locus of management, so sort of understanding who's in control and what they're in control of. Ideally the locus advantage should involve explicit identification of the lead person, the lead clinical manager and management team and their scope of responsibilities. And it's very clear that especially for some of these very complex disorders, the locus of management shifts over time, reflecting the acuity of whatever the individual has, the complexity, and he severity of healthcare needs.

In the paper we described planned co-management, which is the proactive anticipatory approach to care in which the responsibilities are well described and based upon expertise in the capacities in which they work. It involves all sorts of structures and tools, like care plans, formal processes, so how communications happens, and active input of all the key stakeholders, including families, and the affected individuals as the affected individual gets older.

These things often require dedicated personnel who are responsible for carrying out this co-management. And, again, one of the things that we highlight throughout the manuscript is the importance of endorsement of whatever is going on and active engagement of families.

We talk about healthcare transitions, so there are many different healthcare transitions that these individuals go through from short-term follow-up to long-term follow-up as the diagnoses are made, from hospitalizations in and out of hospitals. And then of course what we all strive for, which is a smooth and effective transition from childcare to the adult healthcare system.

In the manuscript, there are a number of specifically really high-level recommendations, which I will summarize as, first, the need to identify innovative programs, including care planning and tools that address co-management. I know a lot of this work is being done in the original collaboratives. The need to incorporate care planning, co-management of family access

functions into electronic health record systems as they develop, and of course all the other related information systems. The need to systematically evaluate preferences, concerns, and needs to make sure that the care which is delivered is really what the individual and the family both want and need. The need to promote outcomes-based research. The need for training to be able to reach what, you know, arguably is an aspirational model that we've been talking about. And, of course, the need to develop methods to incentivize medical home services because if you want to provide really good medical home services to individuals and families, you know, it doesn't come free, but certainly there are a lot of potential benefits that could be gained from that.

So before I open things up to questions, the manuscript has been completed. It was sent to Genetics and Medicine. They had a few little revisions that they've requested, but I anticipate that once Carl returns from the great State of Hawaii, we'll turn that around quickly.

So with that, I'd like to answer any questions you might have.

DR. CHEN: This is Freddie. I've got a question.

DR. KEMPER: Who is that, I'm sorry?

UNIDENTIFIED SPEAKER: Freddie.

DR. KEMPER: Oh, Freddie.

CHAIRMAN BOCCHINI: Go ahead, Freddie.

DR. CHEN: Hey, Alex. Oh, there's an echo. So anyway, thank you so much for your great work. I'm very supportive of the manuscript.

And the two issues that I just want to point out and reiterate. One is the importance of payment reform to support these medical home models. I think this manuscript will add to the ongoing calls or those changes, which are really sort of the crux of what needs to happen to promulgate it.

The other piece is really an ongoing HRSA priority both for maternal child health, as well as the other bureaus, which is around interprofessional team-based practice. And many of us in this field understand that you can't care for these individuals

without an interprofessional high-functioning team.

I think that comes across in your work, but I just wanted to highlight that as another issue. Thank you.

DR. KEMPER: Thank you, Freddie, and we certainly agree with those comments.

CHAIRMAN BOCCHINI: Dieter, and then Don, and then Charles.

DR. MATERN: It might be too late for this suggestion. But I would suggest you change the title where you say "individuals with heritable conditions" to "children with heritable conditions." I think there is a significant issue with adult patients who are diagnosed with heritable conditions and who have a problem finding a medical home. It's just been reiterated by an e-mail I just got on a patient in his 50s who's been going through a medical odyssey until he was finally diagnosed with Pompe disease.

So I think it would clarify it here, and then you can write your next paper on adults with heritable disorders.

(Laughter.)

CHAIRMAN BOCCHINI: Don?

DR. BAILEY: I'm glad you're doing this. This is a great next step.

So I've done a lot of reviewing of the literature over the years of family-centered care or family-centered practices, and I know that medical home is just one kind of one piece of that. But I think HRSA, I think starting back with Roe McPherson had been writing about this for many, many years.

And so every meta-analysis and every study that's been published almost always say that family-centered approaches have resulted in better outcomes. But yet almost all the studies show that the implementation of this in practice is not where we'd like it to be. And you mentioned this as aspirational goal.

So is one of the goals of this manuscript to help -- I mean, it's designed to help improve the use of the medical home model, but I think it needs to be couched in this broader context of which you described at the very beginning, which is patient-

centered -- really family-centered care.

And so I know that Freddie said one of the big issues is, of course, reimbursements and other kinds of mechanisms. But that can't be the only reason why we're not moving in that direction. I do think there are some professional training issues and a variety of barriers. And I'm just wondering if you addressed any of that in your paper?

DR. KEMPER: So thank you, Dr. Bailey. I absolutely agree with everything you said. I think that, you know, funding is necessary, but not sufficient. And I know that, like, I'm looking at Dr. Homer over here, who's really done great work around what it takes to carry this off.

We do highlight the need for training and education to get things going, and also just a real reconsideration of the systems that we, you know, have set up and practices. I talk a lot in the practice that I'm in, is that it seems like a lot of the systems are set up for the convenience of the person working there and not necessarily the convenience of the family.

So we do draw out those issues in the paper. Now I don't think that the manuscript itself is going to change things dramatically, but we see this as kind of drawing a line in the sand, and then I hope we're able to use this to build to other more important things, and certainly HRSA as well. And I know Dr. Mann has been very passionate about figuring out these issues.

So, you know, I hope this is to be continued and not the end of the story. But I think Dr. Homer is probably going to expand on that.

DR. HOMER: No, I think those are excellent points. I was going to ask a question of a slightly different direction.

The paper makes a very interesting distinction between the locus management and the medical home. And certainly in our work, as in many other works, and the paper refers to this when you ask families of children with complex special healthcare needs, they often will identify their specialist as the person who they contact most of the time for most health-related issues for that

child.

So I just wondered if you could elaborate a little bit more on how you differentiate the locus of management from the medical home, because it sounded more in that context that the primary care provider was, in a sense, serving as the preventive care specialist. Perhaps that is they were responsible for immunizations and a few other preventive services.

So I'm just interested because that's been, for me, one of the more vexing issues in this arena.

DR. KEMPER: Well, "vexing" is a great word for this because it is really challenging, and a lot of it depends upon the perspective that you come to these things through. And I wish Dr. Cooley were here because he'd have a much more eloquent answer than what I'm about to give you.

But as we were deliberating these issues and considering them, we really thought of the medical home as that place which was going to provide everything, including first contact

services, as well as the preventive services, and those kinds of things.

It was the experience of the people in the room that it was clear that specialists provide excellent care and care beyond what most generalists could do for these very special conditions, but often didn't have the supports in place to provide those preventive services.

And so the way that we evolved, and I see Dr. Ostrander who's probably going to throw me a rescue ring here in a second.

(Laughter.)

DR. KEMPER: But was that for different conditions, over time there's going to be different locus of control, but primary care still had an important role in terms of providing exactly that primary care. And there just needed to be formal communication to understand who was doing what.

Also I'm going to channel Dr. Cooley for a second, which is always dangerous. But he talks a lot about the failings of primary care and where the gaps historically have been, and really considers a

lot of the medical home space were, a re-branding and rethinking of what primary care ought to be. And so I think that's where that distinction came from.

Did I answer your question?

DR. HOMER: Yes.

DR. KEMPER: Okay.

DR. OSTRANDER: I hope I can help out. It's Bob Ostrander, and I'm on the committee, the workgroup as well.

I'm not sure we delineated as clearly in the paper as maybe we could've in retrospect. But what we meant partly by locus of control is that that's one of the tasks of the medical home is to delineate for the patient, the specialist, and the primary care provider for each task, each aspect of that patient's care where the locus of control will be.

And in integrating that with our requirement, in an excellent medical home there would be a care plan. That's what the locus of control is all about. So in the care plan it says

this doctor will be responsible for getting the annual echocardiogram and the lab work and adjusting thyroid medicine, and the primary care doctor will be responsible for immunizations and blood pressure medication.

So with the medical home's job is to do is to make sure that that question is answered and it's not a big open, I don't know who to call problem for the patient, and then put it in a format that all three parts of that triangle have. So that was what I think the message we were trying to convey is the locus of control is -- determining the locus of control is the job of the medical home. The actual locus of control may be any number of places, including the patient or family.

CHAIRMAN BOCCHINI: Thank you. Could we have your name for the record?

DR. OSTRANDER: Oh, I'm sorry.

CHAIRMAN BOCCHINI: That's all right.

DR. OSTRANDER: Robert Ostrander, and I'm a member of the Follow-up and Treatment Subcommittee and the Medical Home Workgroup.

CHAIRMAN BOCCHINI: Thank you very much.

DR. FRIEDENBERG: Hi, Debbie Friedenberg. Within this medical home, though, there has to be some sort of communication or discussion with the payers because what we have been finding is that the payers will not allow a particular physician to order to order a particular test because it doesn't match whatever. And so I think that there needs to be a lot of education and discussion with the mix of payers as well.

CHAIRMAN BOCCHINI: That's an important point. Yes, Carol?

DR. GREENE: And building on that, occasionally the payers will say that certain kinds of things that are specialized can only be ordered by somebody who really understands them. And that's an argument where you go to locus control and say, oh, let's make sure the person who's qualified to order them does it.

And if referrals -- I'm actually very content, again, as a provider if somebody's got an insurance that says all referrals have to come from

the primary care physician, and the primary care physician is the medical home, and I provide them with the information on what's needed, that's apportionment, and we just have to recognize it. And so building on that.

And I think the manuscript actually talks about the need to talk with insurers and recognizing the insurer as a partner there. That's part of what the medical home does.

CHAIRMAN BOCCHINI: All right. Other questions or comments? If not, do you have the slide of the --

Okay. So what Alex is asking is for the committee to consider and acknowledgment. This would mean that this would not be forwarded to the Secretary, but would then be placed on the Advisory Committee's website with an acknowledgment that basically supports the article.

Can we have a motion to accept that?

DR. BAILEY: So moved.

CHAIRMAN BOCCHINI: Don, Dr. Bailey. Any second?

DR. MATERN: Second.

CHAIRMAN BOCCHINI: Dieter, Dr. Matern. Okay. So we now need, since it's been moved and seconded, a formal vote. And so first we need to know if anybody will abstain from the vote.

(No response.)

CHAIRMAN BOCCHINI: If not, then let's go ahead then and do a roll call vote. We're going to start -- well, we'll start at the top again.

DR. BAILEY: Any proper response format?

CHAIRMAN BOCCHINI: Just if you're in favor, it's yes.

DR. BAILEY: Yes.

CHAIRMAN BOCCHINI: Don Bailey, yes.

Bocchini, yes.

Coleen?

DR. BOYLE: Yes.

CHAIRMAN BOCCHINI: Denise Dougherty?

DR. DOUGHERTY: Yes.

CHAIRMAN BOCCHINI: Charlie Homer?

DR. HOMER: Yes.

CHAIRMAN BOCCHINI: Kellie Kelm?

DR. KELM: Yes.

CHAIRMAN BOCCHINI: Michael Lu?

DR. LU: Yes.

CHAIRMAN BOCCHINI: Steve McDonough?

DR. MCDONOUGH: Yes.

CHAIRMAN BOCCHINI: Dieter Matern?

DR. MATERN: Yes.

CHAIRMAN BOCCHINI: Melissa Parisi?

DR. PARISI: Yes.

CHAIRMAN BOCCHINI: Alexis Thompson?

DR. THOMPSON: Yes.

CHAIRMAN BOCCHINI: And Andrea Williams?

MS. WILLIAMS: Yes.

CHAIRMAN BOCCHINI: All right. It's unanimous.

DR. KEMPER: Thank you very much. I think next time we'll try to be more controversial.

CHAIRMAN BOCCHINI: Okay. Well, you may this afternoon. We'll try this afternoon.

DR. KEMPER: Thank you.

CHAIRMAN BOCCHINI: Next on the agenda is another manuscript, medical foods manuscript, again,

from the Follow-up and Treatment Subcommittee. Sue Berry, subcommittee member will present. And Dr. Berry is professor of pediatrics and genetics, cell biology and development at the University of Minnesota. Welcome.

DR. BERRY: Thank you. Gosh, you didn't get to hear my jokes. All right.

I'm actually quite delighted, all kidding aside, to have the opportunity to share the culmination of a lot of work undertaken through the subcommittee and implemented through the regional collaboratives to gain information about insurance coverage of medical foods for treatment of inherited metabolic disorders.

Like Alex, we'll at the end be asking for committee acknowledgement of this effort. But I wanted to take you through a little bit of where we went with this and what we learned.

Well of course, I think it's worthwhile to think about, as we often do, why we undertook this in the first place. And in one of the things that we had in our survey that I'll describe in a little

more detail, we had a place for comments. And this was a very long comment that I got from one of the parents where her request was to please pass this on. Please pass on her appeal for the desperate straits that she felt herself to be in getting the specialized medical food product that she needed for her child who has fatty acid oxidation disorder.

So I am not going to read this out loud to you, but I would like you to take a look at it when you get a chance when you review those slides because I think this was very emblematic of what we experienced.

So what was the problem that we were trying to encompass with our evaluation here? Well, as most of you are familiar, the treatment for many of the inherited metabolic disorders are medical foods. Some people call it -- the families will call them special formulas. I have one adult woman that refers to it as my milk. They're protein substitutes, but they're not drugs. They're substances of nutritional value. They're not optional. They are the treatment. You don't get to

choose whether you're going to build your muscles by taking it. You need it or you won't live.

Treatment is lifelong in most circumstances unless you just can't afford it anymore. Then it doesn't continue. And everybody needs food, but traditional foods can be harmful to persons with inherited metabolic diseases.

The problem comes in that these medical foods collectively are substantially more expensive than traditional foods, but they may be the only option usable by the family.

Unfortunately in many cases, insurers have taken the opportunity to exclude medical foods from coverage, and that was what we were trying to get a better handle on.

The costs may be quite high. We had to get a little handle on that, and it wasn't easy. In fact, we didn't succeed to all degrees. I can tell you what problems we ran into with that. But coverage is at best variable. And the big issue is that affected persons can't survive without medical foods, but they can't afford to buy them.

So a definition. The FDA regulates the use of medical foods through a specific action in the Orphan Drug Act, and subsequent amendments describes medical foods as foods that are formulated to be consumed or administered internally. This is a point of contention also of whether you think of internally as through your mouth or through a tube seems to be a point of contention. Under the supervision of a physician, intended for the specific dietary management of a disease or condition which has distinct nutritional requirements based on scientific principles established by medical evaluation.

In part, the difficulties rise from this definition alone, and that's a separate problem.

So what are the nutritional treatments just to outline the nature of the character of the treatments. We mentioned medical foods. These are the specially compounded formulas. In many cases it's a substantial portion of nutrition or all of the nutrition for the treatment of the individual.

We also characterized the use of

supplements -- pharmacologic doses of co-factors, vitamins. Biotin for biotinidase deficiency would be an example of that. Amino acids that are required. An example would be citraline needed in urea cycle disorders. In some cases, you get secondary deficiencies and may need a vitamin-like drug like carnitine. And MCT oil, which was, of course, the problem for the lady that gave us her initial plea.

There are also specially manufactured modified low protein foods, which are quite expensive and provide nutritional variety for persons in protein restricted dietary situations.

And then the point that I need to make here is all of these medical foods require physician supervision or healthcare provider supervision. And they're all essential elements of the therapy for the treatments of these conditions. Medical equipment and supplies can also be needed for feeding. That's also an area of impact.

So the Follow-up and Treatment Subcommittee undertook a two-prong investigation of

this. One is we had a group, and it's been a little while now since we did this, where we brought together insurers and a whole spectrum of experts to try and get a sense of what insurance coverage was. And it was almost discouraging because there's such a plethora of different ways of handling it and different exceptions.

Nonetheless, we realize that that mean that there were some very fundamental issues ahead to be addressed with regard to coverage.

We also didn't really have a real sense -- everybody said it was not working very well, but we didn't have any real numbers to prove that. So we said, well, why ask the people who it impacts? We'll ask the families.

We've already alluded to the fact that perhaps some of these things are better characterized as things for children and adults. We didn't even touch the adults in this. And if anything, their problems are much worse. I'm just going to put that out there.

All right. So we had that meeting. And

what are some of the barriers we learned from the meeting? Well, each insurer has their own practices. The private insurers are public insurers using private vendors, self-insured employers, and the practices vary from state to state. Every policy even with the same company can have different coverage, and contracts may result for differences in the same insurer. Every state has its own rules and laws covering provision of medical foods. And this link is still good, but probably requires ongoing updating.

When laws exist, they may not cover all insurance carriers, for example, the ERISA exception. And even when the law and guidelines exist, they're subject to interpretations by the insurers and the States, so there's a lot of capriciousness to this.

So our medical foods survey was to survey parents of children birth to 18 with metabolic conditions to look at their current insurance coverage and actual coverage for medical foods and the related materials. We wanted to have the

opportunity to share this information with policymakers to see if we could reduce some of these financial barriers. And we wanted to know what are the needs that the children have for these products, what are their out of pocket expenses, and what proportion of expenses are paid for.

So we established an expert panel. You can see everybody that knows something about it, including parents, participated in this. We had some initial cognitive interviews because we had to develop our instrument and did some pre-testing of liability planning in the fall of 2008 at three sites, and then undertook the actual survey. And we asked about the diagnosis, the healthcare plan that was covering the child, the products that were used, the extent to which these were covered by their plan, including dollar amounts per month to the best we could get it. Their monthly out-of-pocket expenses if not covered, and if their health plan had caps. And we got nowhere with the caps, so I'm just going to tell you we don't know anything about that. That was hard, but we tried.

So this survey was implemented through the regional genetics collaboratives, and the three collaboratives that participated were the NYMAC, SERC, and the Region 4 Genetics Collaborative. We did this in the summer of 2009, and we targeted it at children.

It was a convenience survey. So it was just whoever walked into the clinic during that month, and we said, will you fill out our survey for us, and they filled it out and gave it back to us in an envelope, so we didn't know who they were. And there were 305 families that agreed to give us that information to the best they could. We had an IRB process. It was a paper survey. It was an anonymous survey. We did not for that reason have a lot of details, other than the state of residence, the age of the child, and what their diagnosis was.

We gave the data back to each of the RCs so that they could use it for their own planning, and then we shared it up for integration in a giant spreadsheet, which is quite intricate.

Just to acknowledge -- I'm not going to

read all these, but I just want to thank my colleagues who were willing to implement this in the various regions.

And this is the heart of the data that was presented in the publication that we've prepared. The first one was the degree to which self-payment was a very substantial fraction of the payment source that was used.

You can see that there is a really good in some ways -- you can look at this as glass half empty or glass half full. Medical foods were covered by a lot of different payers, but they're still a decent fraction of this where the self-pay is a big contribution. And this is particularly egregious for modified low protein foods where 60 percent of families had to pay substantial amounts out-of-pocket.

Dietary supplements, even though they're very drug-like, still were frequently not paid for. And I was shocked that feeding supplies were so often required to be paid out-of-pocket. It seems to me like that kind of equipment should just be

paid for by insurance, and it's totally not. So I was actually surprised by that degree of poor payment.

We were also surprised to find out how much families paid. Again, the glass full here is that there were a lot of people who paid nothing per month for their children's medical foods, and that's good to know. But particularly for modified low protein foods, some families paid more than $500 a month in a visible bar, which I found kind of shocking. And in many cases, families don't have this kind of extra money to be able to pay for these special products, but they have essentially no choice.

So what did we learn? Nearly all of these children had some type of healthcare coverage. That was very encouraging. It turns out that the one or two that didn't were people who chose not to have healthcare coverage because of their cultural background. So that's a good thing. But even if they had them, it didn't necessarily for them. Most children needed more than one category of these

things, and many of them needed all.

Coverage was variable, but there were at least some out-of-pocket expenses for about 20 percent of families using those medical foods, for 30 percent of the families using supplements, for 35 percent of the families using feeding supplies, and about 60 percent of families using modified low protein foods. Those just don't get paid for.

Well, that's something you can keep looking at for a while.

Okay. What do we know and what we don't know. Families didn't know about the caps on their insurance despite the fact that you'd think since they're paying insurance all the time, they kind of, I think, put their head in the sand and don't know this. So we couldn't get this information.

They had a hard time telling us the out-of-pocket costs because they quit keeping track of it. They just pay for stuff and they don't think about it. So it was a hard thing for them to do. We had to go back to some of them and say, can you really kind of help us a little more with this.

The need-based supports are currently very significant. WIC is a really important source of support. And if families were on Medicare ironically, they were a lot better off than a family that was not because often that would be paid for.

Modified low protein foods, I think I've reiterated this. They're particularly poorly supported, and though patterns of coverage varied from region to region, all of the regions experienced -- observed significant challenges. I thought maybe some States would do better, some would not. I will express the caveat that there are States where this is paid for as part of newborn screening programs because it's State-to-State. And we did not engage any States where that was the case. It wasn't because we were picking or choosing. It just didn't happen. And so I want to make clear that the data really reflects States where that is not part of how the medical foods are supplied.

So what's happened? We've shared bits and pieces of this information previously, and the

committee has responded very positively with inquiries to the Secretary regarding medical foods. Two letters have been prepared previously. But in both cases, essentially the Secretary was unable to respond in any way that said, you know, I'm going to do something about this because honestly, there really wasn't some way for that to happen.

One of the problems that we've run up against, and we've talked about this previously, is that we hoped this would be a part of the essential health benefits, and it has not happened. And this is a very big problem in our view, so I'm just going to mention that as a potential conflict, particularly if all of a sudden the benefit package gets set, and State-by-State we have to go back and say, excuse me, don't you want to pay for these things? And that looks like what we may have to do.

Well, I don't want to end on a downer note. I really feel that we learned a tremendous amount from this. And I think this committee has verbalized on multiple occasions, and this is one more time that I'm going to ask you to do that, to

support for families with regard to support of medical foods.

And so what we're asking for you today is committee action to support this manuscript, which was the hard work of a dedicated committee putting together this information and all of the collaborators we had from the regional collaborative activities. We're asking for acknowledgement, and hope that you'll be able to support this. It's been submitted to the *Journal of Pediatrics* and is in review currently.

And I'm happy to answer any questions about this.

CHAIRMAN BOCCHINI: Thank you, Sue, for this excellent discussion on also recognizing the role of collaboratives in providing the resources for you to help make this happen.

Let's go ahead and comments? Michael?

DR. LU: So, Sue, do you know where they are at the IOM? Was that a draft report that came out that said they were going to treat metabolic stuff as -- they didn't say it either needed or

didn't need that essential health benefit requirement, but that it needed another study done.

DR. BERRY: Yeah, and I'm not sure what other study they're exactly looking for. So, no, we don’t have any clear answers from that report.

DR. LU: Was that a final report? I know they got a lot of letters.

DR. BERRY: I don't know what the status of that is at this time, Mike, sorry. If anybody else knows, you should stand up say.

I also want to acknowledge the Newborn Screening Resource Center, who also supported this activity. I just want to thank Brad for making that possible as well.

CHAIRMAN BOCCHINI: All right, Steve? Oh.

DR. CAMP: Oh, hi, thank you. Cathy Camp. With respect to the IOM report, that had been -- the draft was end of last year. And it referenced medical foods as considered to be exempt. However, that has gone back to the Secretary, and that whole package has now gone back to the States.

So in the Affordable Healthcare Act --

this is my understanding, and if anybody knows any different, pipe up. But the definition or the establishment of a "package" will now be back in the hands of the States.

DR. BERRY: And that was what -- yeah, I think that that's part of our sort of --

CHAIRMAN BOCCHINI: Concern.

DR. BERRY: -- feeling that we're going to have a real hard time establishing that State-by-State. Steve, did you have --

DR. MCDONOUGH: It's a great thing that you've done, and it'll be nice to see that in pediatrics.

This is basically a political issue from my perspective, and part of the political process where some things are recommended to be covered despite tremendous controversy, which are very worthwhile. And others are quietly not covered. And hopefully this will be revisited in the future, this issue.

I have one question. Did you see an age difference? In my State, all children I think would

get Medicaid and WIC coverage as far as insurance because they'd be eligible for early childhood development. But by age three, they're kind of on their own and they're SOL. So I don't know if you saw that, if the teenagers are having more troubles than the early kids.

DR. BERRY: We weren't able to do that kind of detailed analysis. But I would say that it was clear that WIC was an important contributor when it was supplied. And we agree that that means that we miss a lot of people who aren't WIC eligible.

And, again, we didn't even try to look at adults anecdotally. Some of the people gathered surveys from adults, but we didn't include them in the analysis. We didn't have enough, and we weren't trying to look at that. But it didn't look good in what we had, the stuff we received.

So one of the things that would be probably appropriate to do would be to go back and look more carefully at young adults and persons in transition, and then adults who are being cared for who require medical foods.

CHAIRMAN BOCCHINI: Dieter and then we'll go back --

DR. MATERN: Sue, do you know is there any data about the costs that would incur to the health insurance companies if they had to pay for all this?

DR. BERRY: You mean the total costs?

DR. MATERN: Yeah, compared to everything else, I mean, this is peanuts.

DR. BERRY: This is a drop in the proverbial bucket. Really the cost analysis that have been undertaken, this is really a very small fraction of healthcare costs, which makes it sort of ironic.

CHAIRMAN BOCCHINI: At the microphone.

DR. MORRISON: Thank you for this report. My name is Bill Morrison. I'm a parent advocate with the Taskforce for Genetic Alliance. I'm also a nurse, and I can tell you that from my experience working with the special needs community, which I have been doing for 15 years, that, like Dr. Dieter mentioned, there's a drop off point with a lot of the resources for these people. And I worry about

that.

My son is 13 years old. He has PKU. Luckily he's been covered for his medical foods under my insurance program. The low protein foods have not, and it's a very significant cos. But I worry about him as he becomes older and becomes his own person, and has to move into his own insurance coverage, and can't be covered by my policy anymore. What's going to happen to him and what is the effect of the fact that he's healthy. And, you know, we just had this assumption that you become an adult, you know, you've got the problem covered.

And so I would encourage this body to stay on top of this, and please continue to revisit this because, you know, a lot of these kids are going to become adults very soon. And my son is only six, seven years out. And what's going to happen to him? You know, it's a big concern for me as a parent, and I'm sure all parents experience that with their children.

And it is not a huge, you know, cost to the insurance companies. Something needs to be

done. You know, these are rare disorders. The kids aren't astronomical in numbers, you know. I think on our insurance policy with our private insurance, our son's the only one. You know, so they can help with it. They've just got to be told by somebody with authority. Thank you.

CHAIRMAN BOCCHINI: Thank you.

MS. HUNTINGTON: My name is Kathleen Huntington. I'm a clinical dietician from Portland, Oregon, and I'm a co-author of another published paper on medical foods and the lack of insurance coverage.

I want to give you some perspective on terms of total cost and it how it impacts on total healthcare dollars.

The Council on Affordable Healthcare analyzed this, and they're an organization that looks at the market remedies for healthcare costs. And they deduced that it was less than one percent of total healthcare dollars. We calculated that it was actually one-fifty-fourth thousandth of total healthcare dollars.

UNIDENTIFIED SPEAKER: Rounding up.

(Laughter.)

MS. HUNTINGTON: So it's really important -- it's actually 28 States that have passed in their mandates coverage for low protein modified foods -- modified low protein foods. So the majority of the States, out of 38 who have passed mandates for coverage, the majority of them have included medical modified foods.

Well, what's important about this is that that reduces the total healthcare costs for these treatments because if you use only formula to meet calorie needs as well as protein needs, it's much more expensive. But if you use a combination of the medical protein formulas and the modified low pro foods, it's much more realistic for insurance companies since they have to cover this, as well as the families, because the co-pay is rather large and generous.

And another thing to point out is that WIC only covers to five years of age, so it's a very important assistance in State covered dollars in

terms of Medicaid to use WIC. It only goes to five.

DR. BERRY: Common sense should reign, but I don't know if it will.

MS. MONACO: Hi, I'm Jana Monaco with the Organic Academia Association. And as many of you know, I have two children with one of these disorders, and I can attest to everything that Sue has given in her report.

And I wanted to just hone in on -- we actually do live this, and it's been almost 11 years since our son had his metabolic acidosis, and we became dependent on these formulas. And he is strictly fed through his G-tube. But then my daughter came along, and when she was first born, we were denied covered because she took it by mouth. And actually it was Dr. Green who had to write a wonderful letter to our insurance company when she was her care provider stressing that this was needed and important for her.

Living in the Northern Virginia area, my husband is one of those beltway bandits and government contracting to self-insured companies.

And so we have different experiences over the 11 years as far as full coverage and not full coverage. And my concern in the way this has occurred with the legislation that is kind of sitting on someone's desk, this effort to -- with the IOM and everything, and now being put back to the States. It's such a pass the buck situation, and no one seems to want to own this problem, but leaving it really back to the families because in the State of Virginia, after we got expanded screening, we went after the formula coverage issue. And it was kind of determined that let's look at the Federal level and see what's happening there.

And at best in our State right now, the only way a family can get any free formula, per se, is if they fall 300 percent below the poverty level. We don't fall into that current category. And then you have the self-insured issue.

So when, for instance, my husband took his job four years ago, there was no coverage, so it was $1,700 a month out-of-pocket for two children, and that included the formulas, the pumps, the supplies,

the actual G-tube buttons, et cetera. Now we have -- our son, Steven, is provided by Medicaid, but our daughter is not. So we are paying half of her formula costs.

And in an effort to get her to eat more foods, we ran into a severe problem with this and showing that she can't do without it. We had to actually increase her formula recently, so it's very important. The supplements, that's another extra hundred dollars a month. And even that has not been recognized, and we've attempted that in 11 years, too.

So it is a real problem, and I don't accept the fact as a parent that the Secretary should be excused from HRSA to provide no further action. I think somebody has to continue to address this issue. And if left to the States, a lot of States have discontinued their programs to fund these formulas. And really they're looking for this. And this unfortunately is going to be an out for some of the States to find more reasons why they can't pay for it. So I hope you will continue to

keep this on the table. Thank you.

CHAIRMAN BOCCHINI: Thank you for your comments. I think it puts a personal feel to the issues.

So we'll take one more comment, and then we'll look by the committee to go forward.

MR. PENN: I'm Jeremy Penn. I had a quick question about the FSA and HSA, if they're options for families to help them afford medical foods, or if those are not allowable expenditures in those categories.

DR. BERRY: I think in many cases families can use whatever resources they can dig up, and so sometimes those are possibilities. But, you know, it shouldn't be a piecemeal event. It shouldn't be when I have this insurance I get this, and when I have that insurance I get that. It's just so hard for families. I think you heard from Jana extremely eloquently that even if one circumstance that's possible for another, it won't be.

I have a woman that is an extraordinarily dedicated care coordinator for our newborn screening

program, who spends about a third of her time writing letters of appeal for this stuff. Talk about waste of time. I could certainly use her intellect and abilities doing more productive things than writing still another appeal letter that's going to be turned down.

We've gotten to the point where whenever you ask for a specific supplement, you just put it through so that it can be denied so you can write the letter. We have epic templates for appeals. That's sad. Sad, sad, sad, yes.

CHAIRMAN BOCCHINI: Don?

DR. BAILEY: So is this a problem that someone could own, or is it the problem that really -- and we just don't have access because we're in the Secretary -- we're in HHES, or is it no one really owns this problem?

DR. BERRY: I think that Jana kind of told you what the situation was, which is you could do it as a Federal mandate if you had either a legislative solution or if you created an expected benefit package that was uniform. But clearly that is not

the direction that it came with, and so now that it's passed back to the States, it's going to be chaos.

CHAIRMAN BOCCHINI: So on the table today is the consideration of this article to go forward, and a request for committee acknowledgment. I will entertain a motion to do so.

DR. MCDONOUGH: So moved.

CHAIRMAN BOCCHINI: Steve, so moved, Dr. McDonough.

DR. PARISI: Second.

CHAIRMAN BOCCHINI: And, Dr. Parisi, second.

So now we'll have to have a formal vote. So this time, I think I'll start with Dr. McDonough, and then we'll go reverse alphabetical order.

DR. MCDONOUGH: Aye.

CHAIRMAN BOCCHINI: Aye, okay.

(Laughter.)

CHAIRMAN BOCCHINI: Dr. Lu?

DR. LU: Yes.

CHAIRMAN BOCCHINI: Kellie Kelm?

DR. KELM: Yes.
CHAIRMAN BOCCHINI: Charlie Homer?
DR. HOMER: Yes.
CHAIRMAN BOCCHINI: Denise Dougherty?
DR. DOUGHERTY: Yes.
CHAIRMAN BOCCHINI: Coleen Boyle?
DR. BOYLE: Yes.
CHAIRMAN BOCCHINI: I will vote yes.
Don Bailey?
DR. BAILEY: Yes.
CHAIRMAN BOCCHINI: Andrea Williams?
MS. WILLIAMS: Yes.
CHAIRMAN BOCCHINI: Alexis Thompson?
DR. THOMPSON: Yes.
CHAIRMAN BOCCHINI: And Melissa Parisi?
DR. PARISI: Yes.
CHAIRMAN BOCCHINI: Dieter Matern?
DR. MATERN: Yes.
CHAIRMAN BOCCHINI: Okay. All right, thank you very much. Thank you, Sue, very much for that presentation and leading the discussion.
All right. So now we have a presentation

by Meredith Weaver. Dr. Weaver is a Board certified genetic counselor and associate project manager at the American College of Medical Genetics and Genomics. And today she's going to present an update on the project of the Carrier Screening Taskforce and its interval findings. Welcome.

DR. WEAVER: Okay. Hi. Thanks for having me. Since we're right before lunch, I'm just going to go in without my slides. Hopefully they're coming soon.

So I'm just going to tell you the purpose of the workgroup, who is on the workgroup, the timeline, what we've been working on over the past year and a half, the highlights of the progress, and then what our next steps are, which we're hoping to get a little feedback from the Advisory Committee.

So the purpose -- I do have my notes in front of me. I apologize you guys don't have it. There was a charge from the Secretary Advisory Committee in the summer of 2010 -- so about a year a half ago -- to develop a workgroup on population-based carrier screening. And the actual charge was

to engage a multidisciplinary stakeholder group using the modified Delphi process -- and I'll talk about that a little bit -- to collect and document perspectives on public health, personal health, and healthcare system readiness and needs -- public health, personal health, and healthcare system readiness and needs for expanded population-based carrier screening for genetic conditions. Remember this was back in the summer of 2010 when population-based carrier screening was in the news. It was being talked about a lot.

So the end product is to be a report to the Secretary Advisory Committee that will include an outline of recommendations and a roadmap of considerations that are needed prior to implementation of population-based carrier screening.

So we convened a workgroup that includes all these different types of professionals. So we have ethicists on the workgroup, legal experts, healthcare providers, advocates, consumers, and representatives from commercial labs. And obviously

some people on the workgroup actually wear more than one hate.

Here's a list of the workgroup members. Some of the people actually are in the room today, and the co-leaders you can see down at the bottom are Sara Copeland and myself.

So here's our timeline, what we've been up to for the past year and a half. In September we were given the charge to create the workgroup. We had our first in-person meeting in January of 2011, and at that point, that's when we talked about what topics needed to be surveyed about the topic population-based carrier screening.

So after that in-person meeting, the taskforce was organized into a workgroup. It was formalized by Rod Howell and Michelle Puryear. And based on what we talked about in January, we developed a Delphi survey, which was pilot tested in March of last year.

In April we had the first round covering both May and June. We had the second round, and I'll mention this again, but the Delphi survey is --

it's iterative. It's rounds of questionnaires. And usually there's three rounds, but we stopped after two rounds because there was a saturation of opinion. There was nothing new that we were hearing.

So last summer the workgroup as a whole was divided into breakout groups to review the primary data, and the breakout groups mirrored the topic areas of the survey.

In September, the breakout group members reviewed the primary data, and so they looked it over to see what the responses were and kind of what points were hit upon. In November of last year, we had a conference call of the whole workgroup to discuss the progress. Okay, what did you see in the primary data, and what do we need to do next?

So in January of this year, the breakout groups had the unenviable task of summarizing those findings from the primary data. And then in April of this year, we asked the breakout group members for their own recommendations in terms of population-based carrier screening. Okay, given,

that you do what you do, whatever those professional groups you fit into, what, in your experience, is important for people to know before population-based carrier screening is implemented? What do your experience and your expertise tell you?

So currently that's May of 2012. We're revising and collating the summaries and the recommendations. And then looking forward this summer, we're going to do revisions to that report and anticipate submitting the report to the Advisory Committee in September.

And then again, this is kind of in anticipation of October of this year, sending the report out for public comment. It'll come back to us in November and December, and we'll revise it and finalize it with the ultimate end goal of January 2013 having a final report presented to the Advisory Committee.

So what have we done? We started with literature review when the charge first came through at the end of 2010, and this was updated by Don Bailey in September of 2011. He added more search

terms and added in anything that had been published in 2011.

The modified policy Delphi, that was the two rounds of questionnaires that I mentioned. It's a mechanism that's used to develop reports. It's a way to gather diverse opinions from experts on a topic area, then to make recommendations to something like the Secretary's Advisory Committee or the Secretary of Health and Human Services.

The important word here is "modified policy Delphi." The policy Delphi is a mechanism where people come to consensus around a topic. Modified means that we're actually looking for areas of non-consensus, so where people disagree about population-based carrier screening.

I mentioned earlier that we only had two rounds because we were getting saturation of opinion. So what we found in our two rounds were several areas of non-consensus, and I've broken it out into the five topic areas. The first topic area was social issues, so people did not agree on how to release the ownership of results, the access to

results, and the storage of carrier test results. So there's differing opinions about those four issues.

In terms of psychological issues, whether it was feasible to consider the psychological implications and individual life experiences. These are complex issues, and obviously some respondents thought it was not feasible.

In terms of economic issues, the scope, purpose, and desirability of a cost-effectiveness analysis, again, a complex issue. How much to look at, what would be the purpose of doing a cost-effective analysis?

So the fourth topic area is the education and communication issues. And then the non-consensus was whether it was feasible to consider shared decision making. Again, this is a complex issue, and how is this to be done on a population-based level with the results coming back, and whether it's feasible to provide comprehensive genetic counseling. This was more around does everyone need comprehensive genetic counseling. How

should it be done? Is it in person? Is it web-based?

And the last topic area was testing issues. And this is how to report secondary information to the patient, the evolving information base. So when new information about natural history comes up, what to do about that, the duty of the provider to inform or re-contact when new information becomes available. And there was non-consensus about potentially or eventually using whole genome sequencing as the screening method, how that would look.

So I said there were several areas of non-consensus, but the vast majority of the issues had consensus. So these are the highlights again. This isn't comprehensive, some examples. So informed consent should be required. Everyone agreed. The feasibility -- it's feasible to consider the burden of carrier screening to the healthcare delivery system. It's important to consider the cost of follow-up testing or procedures or actions. It's desirable to have shared decision making. Two

slides ago I said there was non-consensus about whether it was feasible, but everyone agreed that it was definitely desirable. It's desirable to have kits and reagents widely available, talking in terms of IP issues, intellectual property issues. And it's feasible to have comprehensive science and empirical evidence available to determine clinical utilities.

So these are just some of the highlights of what the respondents agreed were important.

So this is kind of the big slide. So the anticipated Advisory Committee actions. In September, to review the report that comes out of this workgroup, to make comments. Then we would like recommendations for eliciting input from relevant populations. So this is our public comment period. How do we go about doing that? Who is going to be giving us comments back? And then to help us determine the final disposition of the report. So what does it look like, who does it go to in January of 2013.

And I spoke really fast. We're going to

lunch.

CHAIRMAN BOCCHINI: Thank you, Meredith. That was a great summary of the progress that you've made so far in the project.

So any questions, comments from the committee? Steve?

DR. MCDONOUGH: A question. What are the options to acknowledge like we've done this morning to send on to the Secretary? What other options do we have for the report?

CHAIRMAN BOCCHINI: Well, I think all options are open, but obviously we'll wait as this evolves. And in January when we have the final report, we will then make a determination at that point as to what to do and see where this best fits.

Additional comments?

(No response.)

CHAIRMAN BOCCHINI: If not, thank you very much. I think that this seems to be progressing quite nicely, and we look forward to your final or next interim report. Thank you.

All right. Well, now we've worked our way

to the lunch break. I know we're a few minutes late, but to stay on track for this afternoon to get people who have flights out of here on time, I think it's best that we reconvene at 1:00. And so that'll shorten the lunch period by 15 minutes, but if we can all make it back by 1:00 p.m., we'll get started on schedule. Thank you.

(Lunch recess.)