U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

HUMAN RESOURCES AND SERVICES ADMINISTRATION

DISCRETIONARY ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN

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MEETING

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FRIDAY
SEPTEMBER 12, 2014

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The Meeting convened via webinar, at 9:00 a.m., Joseph Bocchini, Chairman, presiding.

PRESENT

JOSEPH BOCCHINI, Chairman, MD
DEBORAH GOLANT BADAWI, MD
DON BAILEY, PhD, MEd
NATASHA F. BONHOMME
JEFFREY BOTKIN, MD, MPH
COLEEN A. BOYLE, PhD
FREDERICK M. CHEN, MD, MPH, FAAFP
SIOBHAN DOLAN, MD, MPH
DENISE DOUGHERTY, PhD
CAROL GREENE, MD
CHARLES F. HOMER, MD, MPH,
KELLIE B. KELM, PhD
FRED LOREY, PhD *
DIETRICH MATERN, MD
STEPHEN McDONOUGH, MD
MICHAEL LU, MD, MPH
MELISSA PARISI, MD, PhD
NANCY ROSE, MD
DEBI SARKAR, MPH
SUSAN TANKSLEY, PhD

BETH TARINI, MD, FAAP
ALEXIS THOMPSON, MD, MPH
CATE WALSH VOCKLEY, CGC
MICHAEL WATSON, PhD, FACMG
CATHERINE WICKLUND, CGC
ANDREA WILLIAMS *

* Present via Teleconference

P-R-O-C-E-E-D-I-N-G-S

10:56 a.m.

CHAIR BOCCHINI: All right, let's call the meeting to order. We've been able to get all the lines open, and we'll go ahead and call the meeting to order. All right. Able to hear? Okay.

Good morning, everyone. Welcome to the second day of our September 2014 Discretionary Advisory Committee on Heritable Disorders in Newborns and Children meeting, and thank you all for being here the second day.

First item of business is attendance. Call the roll.

(Roll Call)

CHAIR BOCCHINI: So, we have another full agenda today, and we're going to start this morning with the report from the Laboratory Procedures and Standards Subcommittee. They're going to provide an update and some draft recommendations for timely newborn screening on the project that they've been working

through. Over to you, Kellie.

DR. KELM: So, thank you for giving us a very generous amount of time this morning to go over what we've been working on since this whole process started about a year ago. So, Susan and I are going to talk about what we've been doing, and propose some recommendations.

So, the slides that we have here have changed somewhat since --- compared to the ones that you have in the briefing book. We had a lot of great discussion last night in our Subcommittee meeting and a lot of changes happened, especially to the revised recommendations that we're going to be presenting today.

The purpose of what we've been working on is to report on best practices to alleviate the gaps and identify barriers to timely newborn screening and assess whether current goals for timely specimen collection and transit and testing are appropriate for the current newborn screening system.

So, if you recall in January at the Committee meeting, the Advisory Committee recommended the following time frames related to newborn screening. And a lot of these came from the 2005 report that has been on the Committee website for over eight years.

So, initial newborn specimens should be collected at 24 to 48 hours of life. Newborn screening specimens should be received at the laboratory within 24 hours of collection. Newborn screen results for time-critical conditions should be available within five days of life, and all newborn screening results should be available within five days of collection.

So, our Subcommittee was tasked with the following six items. First, to outline the newborn screening system in order to inform the rest of the items, to investigate existing gaps and barriers in the newborn screening systems, to identify best practices to achieving these goals, to develop

a list of critical conditions that require urgent follow-up, review the recommendations in light of new technologies, and suggestion revisions, if needed.

So, in order to meet these tasks we convened a steering work group made up of the following Subcommittee members, Stan Berberich, Dieter Matern, Michelle Caggana, Mei Baker, George Dizikes, Bill Slimak, Debi, Tina, Susan, myself, Ed McCabe from March of Dimes, and several staff from APHL who've been fantastic. This whole group has put in --- we've had calls every other week in addition to some of the other work, in addition to the calls that we've been doing on a regular basis, so we want to extend our thanks to this whole team for putting in a lot of time and effort on the work that we've done so far, and I'm sure the work that's going to be ongoing.

So, as I said we developed --- we had biweekly calls and more, and we started by developing an outline of the system, had a

discussion guide, and then using this discussion guide work group members started by holding focus groups at two regional collaborative meetings, and that was sort of the start for what wound up being a survey that we used for the states.

So, working with APHL they conducted a survey of the states. And as I said, we used the focus group results and common themes to guide the development of the survey questions and the answer choices.

We also had discussions on critical conditions with several groups, and we'll do a little bit more discussion of what SIMD has been working on, as well. And we've been having calls with expert groups in the field of endocrinology, pulmonology, hematology, and immunology to capture their thoughts on critical conditions. And we're still in the process of working with the Joint Commission and American Hospital Association to work on our partners in the hospitals that are actually

working collection and shipping, and Dr. Dolan will actually give us a little bit of flavor at the end of our talk on what the March of Dimes has been working on, which I think is complementary to what we've been working on.

So, as I said, the first thing that we wanted to do was to outline the system. And in the upper lefthand corner what this diagram shows is that there are many parts of the newborn screening system. And it's important to involve all of those in the system. And, of course, the baby is in the middle. We're doing all of this for the baby. So, in the lower right-hand corner, this diagram will be --- we talked about really changing it and we just didn't have a chance before today's meeting, but it shows a little bit more of the linear --- somewhat linear process that's happening starting with parent provider education, and then the testing that happens, you know, after the infant is born from specimen collection, transport, receipt at the lab, testing,

results, verification, reporting, and then, obviously, all of the follow-up that happens. And overarching all of this is the constant quality improvement that's happening on every step of this process.

The first thing I'm going to talk about is our work in developing the list of critical conditions that require urgent follow-up. So, in terms of hemoglobinopathies we worked with HRSA to reach out to the experts that were utilized in the case definitions project previously. And at this time, the consensus within that group of experts was that their conditions do not require urgent follow-up, so they're not critical.

The endocrinologists we talked to last week similarly were the endocrinologists we had used previously in the case definitions project, and their decision or recommendation was that CAH was considered time critical. They would like the results within five to seven days. And CH is time sensitive, which means that

although these are not time critical conditions that need to be performed immediately, that all newborn screening for all these conditions is time sensitive and needs to be done as soon as possible. And these results they would like to have available within seven to fourteen days.

CS, we're also using experts utilized in the case definitions project. There's been a lot of communication by email, and we're planning on having a discussion with them next week.

And the last one I'm going to hit on is the metabolic conditions. So, we in our Subcommittee yesterday, Sue Berry came and gave a great presentation on their work. So, SIMD had during a meeting this spring, there was interest from clinicians to work themselves on outlining what they considered the critical list of conditions, so that --- because they often work with the Public Health system, and they felt that that would also be useful. So, it wound up luckily working at a beautiful time

for us to be able to leverage the work that they were doing in their group.

So, they completed --- the work group that they had completed a position statement and that's been provided for us. And I wanted to let you know their definition of a critical condition is presented at the top, so it's condition on the RUSP in which acute symptoms or potentially irreversible damage could develop in the first week of life, and for which early recognition and treatment can reduce risk of morbidity and mortality.

And here is the list of critical conditions from our primary, the list of primary conditions on the RUSP. There are also several critical conditions that are on the secondary --- a secondary sort of list on the RUSP that we didn't capture here but we, obviously, had the information from SIMD and we'll with their permission include it in the work that we're doing in our report.

The position statement further

includes other details on, you know, what they recommend in terms of maintaining appropriate standards of collection. It's important to have presumptive positive results as soon as possible with immediate referral for appropriate evaluation and management.

These conditions can present with potentially lethal crisis in the first hours or days of life, and here are some quotes that we've pulled from their position statement that we felt were important to still draft. It is not possible even the most ideal system to have results of newborn screening available within --- for clinical presentation of all affected babies, and some babies will present even before its proper to collect the newborn screening sample or specimen.

And the clinicians must include inborn error metabolism and the differential diagnosis of an ill newborn. And, finally, be aware of clinical variability. Clinical response may depend on the analyte level,

analyte patterns and ratio of analytes, and there's heterogeneity in the severity of conditions in the spectrum of clinical manifestations. Each condition listed has a significant risk of catastrophic presentation in the first week of life though many babies with a critical condition may be asymptomatic in the first weeks of life. And some babies with conditions on the RUSP that are not included in the critical conditions list may still present in the first week of life.

Now I'm going to pass this along to Susan, and she's going to talk about the survey of states that APHL helped us to ---

DR. TANKSLEY: Good morning. So as Kellie mentioned, our first approach to gathering data was to go to the regional collaboratives. And some focus groups were held at those two regional collaborative meetings, and information was gathered from that. And we used that information to help develop the survey questions.

We wanted to get --- our goal in gathering information was to try to gather information from every state in the nation. It was very important that we received input. We wanted to know what are the specific gaps, what are the specific barriers that you face, what are the things you've done to improve timeliness for collection, for screening, for transit.

So, a survey instrument was developed by APHL and with the work group's assistance. It consisted of 31 questions. There were three different sections of the survey. The first focused on communication between the states and birthing facilities. The second focused on the newborn screening training program, and the four recommendations related to timeliness, so gathering, the gaps, the barriers, and the best practices. And then the third section focused on new technology, new tests, and their impact on timeliness because that was something else that the work group was

to assess.

Are very happy to report that we had tremendous success in gathering information from all the states. And there were three emails sent out to every state, one went to the Public Health Lab Director, one went to the Newborn Screening Lab Director, and one went to the Follow-Up Coordinator. And that was a way to try to insure that we would --- every state at least knew that this survey was out there to be fielded.

There were two different versions of the survey. There was a lab version and a follow-up version. The states had the option of submitting both or submitting a singular one for both. We did ask that if only one survey was to be sent back that the Lab Director survey be sent in, and that's because it included some quantitative data requests; whereas, the follow-up one only include the qualitative ones.

So, we had 62 surveys submitted, 47

of those utilized the Lab Director version, 15 used the Follow-Up Coordinator version, and in total that represented all 50 states and Puerto Rico. So, we were extremely pleased to have gathered information from all of the states. The survey was open from July 8th to the 31st.

All right. So, first we'll talk about the first part of the survey, which focused on communication between state newborn screening programs and birthing facilities. So, what came out in the survey was that all newborn screening programs provide feedback to individual birthing facilities. Some of the feedback includes unsatisfactory specimens, transit time, completion of essential information, and the age at specimen collection. That feedback may be provided monthly, quarterly, or as needed.

Technical assistance or training is provided the birthing facilities by 50 of the 51 programs and it's typically upon request or

upon recognition of an issue that they've been monitoring.

Then a question was asked trying to determine if there's a mechanism to note if all babies in the state are actually screened or not. So, 30 of the states have a mechanism, and some of those mechanisms include matching the newborn screening specimen to vital records or birth certificates. That may happen daily, weekly, or monthly. Some of the states have newborn screening specimen card kit numbers submitted with the birth certificates which links them. I think that was an issue that had come before this Committee a few years ago. So, for the states that aren't able to have this sort of linkage, some of the barriers were that there isn't this linkage between newborn screening records and vital statistics, so the inability to link with vital statistics.

One of the states noted a failure to link directly to Amish populations, to other home deliveries, and babies born out of state.

The availability of birth certificate data at the time of screening is an issue, and also some states noted that there was no way to capture parent refusal.

All right. So, moving on now to survey data in the second part of the survey. This is where --- the first thing we wanted to do, and we asked essentially the same questions for every recommendation. So, we asked basically what is your current status, so how well do you meet the current recommendation? We asked what are your gaps and barriers? And we asked what are your best practices? We also asked what are the three most important things that you think could be done to improve timeliness for that particular recommendation?

So, the first recommendation is initial newborn screening specimens should be collected at 24 to 48 hours of life. Each --- to orient you with these, each of the bars represents a newborn screening program, so you can see the spectrum runs from 11 percent to

98.3 percent that meet this recommendation currently, with the median at 82.2 percent. So, this is being met pretty well right now.

So, which factors impacted? So, you have the full survey with the responses analyzed in your briefing book, so we're just summarizing the data today. But if you haven't already looked at that, you can look at the details.

The states were also asked to rank the impact level for each of the barriers from a major impact down to no impact.

The factors that newborn screening programs rated as having a major impact on their ability to meet the goal, so compliance with collection from premature and sick infants was the highest at 23.5 percent as a major impact. Transfer of newborn before the specimen is collected, release of newborn prior to 24 hours of life, and high turnover of staff performing dry blood spot collection.

Now, I should note at this point if

you recall who we sent the surveys to, the surveys were sent to the state newborn screening program, so this is information from the perspective of the state newborn screening program. And we weren't able in this time frame to survey the birthing facilities themselves; though I know that some of these states have actually contacted birthing facilities and received input such as this.

Other gaps and barriers that were noted, midwifery centers and out of hospital birth, lack of education to submitters and parents due to low staffing, high turnover at birthing facilities, and state regulations that allow collection at different times than the 24 to 48 hours.

Some of the best practices, so between --- this is a compilation of what's currently being done, what could be done. And they matched for the most part. So, many states were already doing the things that were being recognized as the most important things that

could be done.

So, providing education and outreach to individuals involved in newborn screening processes, so this is formal education, provider education, the whole gamut. Monitoring performance and providing feedback and technical assistance to birthing facilities, which as I noted in the very first slide that we presented that that's already being done by all states. And to make legislative changes or revise state regulations to match recommendations and provide regulatory --- these are two separate things. And to provide regulatory authority to insure compliance. So, although there is statutory authority, often there's not regulatory authority.

And there was one state --- so, there was also a free text portion for each survey part, and one of the states noted as a recommendation to make this a Joint Commission standard.

For the second recommendation, newborn screening specimens should be received at the laboratory within 24 hours of collection. Again, we note 31 states responded to this question, and the response is from .6 to 80.8 percent that met this goal at this time, the median being 25 percent of the specimens at this time, or receiving specimens within 24 hours of collection.

For the factors that newborn screening programs rated as having that major impact on their ability to meet that goal, geographic distance, 37.3 percent. The lab not accepting specimens on weekends or holidays, 29.4 percent. Operating hours of the lab and courier both were at 27.5 percent. Lack of a dedicated courier at 25.5 percent, and batching by birthing facilities at 19.6 percent.

Other gaps and barriers noted were more things about courier services and other mail delivery challenges. Birthing facilities, submitter challenges in getting specimens sent

out, which may be another form of batching. And lack of timely feedback to birthing facilities and submitters on performance.

So, as far as best practices are concerned for meeting this goal, utilize courier and/or overnight delivery services so mail, U.S. mail was still utilized quite a bit for newborn screening. Provide educational activities to birthing facility staff, laboratory staff, and parents. Continuous quality improvement activities with birthing facilities and submitters. Performance monitoring and feedback, and expanding newborn screening lab operating hours.

We were also tasked, as Kellie said, with coming up with the list or developing a list of time-critical conditions. So, we wanted to find out how many states already have a list of time-critical conditions. So, these are conditions --- and the way we defined it for the states was you do something differently with these. You know, you're reporting them out as

quickly as possible. You may be doing something different. Maybe they're reported out before they're final, if you have a gap between when it's going to be final and now. So, 37 of the states said they already had conditions that they considered time-critical, 14 states did not.

A list was --- and it's available in your briefing book, but a list was gathered, so we took a draft list from SIMD and sent it out as part of the survey basically just asking does your state currently consider these to be time-critical? And those --- that's --- all the details are listed in your briefing book so that you can see what's already being done. But there are also many other conditions that are already also considered time-critical in some states.

So, the question --- the third recommendation, that newborn screening results for time-critical conditions should be available within five days of life. So, when we

asked states what their current status was, we had 17 states that responded, and the range was from zero percent to 99 percent, with a median of 75.8 percent.

So, I do want to make a note because you've seen that the numbers have dropped off on the amount of quantitative data that's coming back. And one of the things that we note and something that everyone needs to be aware of is that we had that short window for the collection of the data. So, laboratory information management systems, you have to have the data in a form that you can pull it out, and you have to have the ability to pull that data out in order to respond to a question like this. Many states responded that at this time they weren't able to pull that data, so they may have had to contact a vendor to get a new query built which would take longer than the amount of time we had for the survey.

So, I also want to note that I talked with Marci Sontag last night, and NewSTEPs is

in talks with the LIMS vendors so that the quality indicators that have already been proposed for the NewSTEP website, they're in talks with them to get those built into the LIMS for the major vendors. So, she volunteered that they could also talk to the LIMS vendors about putting whatever the final determinations are, putting those into the LIMS so that that data would be easily queriable for the states, as well.

So, the major factors impacting states' ability to meet this goal, specimen receipt time falls outside the recommended time frame. So, specimens are received at five days of life, you have no chance of getting the results out in five days of life.

The operating hours of the courier, operating hours of the lab, the lab not accepting specimens on the weekends or holidays, some home births are not reported. And then in some instances second tier testing impacts the ability to turn around those

critical results. So, if you have another test built onto the end to try to reduce the false positive rate, then that impacts your ability to get the results out in a faster manner. So, we have to figure out a balance, as well, so that we don't increase false positive rates because we're trying to get results out faster. So, it's another thing to consider as we look at this.

Also noted was use of an out of state laboratory, and that does happen --- two of the 37 states responded as that, but there are more states than that that utilize out of state labs.

As far as best practices, providing education. You've heard it on every recommendation. You'll continue to hear it. We think it's a very important issue. Increasing newborn screening program operating hours for both the lab and follow-up, so the ability to report out the results, make sure those results are received by a healthcare provider. Providing courier or overnight delivery services, or encouraging their use. Monitoring

performance and providing feedback to birthing facilities, couriers, and laboratories, and focusing on newborn screening program improvement, such as increasing capacity, decreasing turnaround time, use of technology.

And then the final recommendation was that all newborn screening results should be available within five days of collection. So, the --- let's see, 22 states provided data for this, and again zero percent to 100 percent met this goal, with the median being 81.9 percent.

Factors that had a major impact, delays in the processes. So, basically, all the things we talked about before, so that we didn't have to reiterate them. Operating hours of the lab, the test itself. So, what that means is the test doesn't take an hour, it takes a day, or a day and a half to complete. And as we add more and more testing, and more and more complicated testing, that does impact turnaround turn within the laboratory.

Noted was the ability to implement change, so although you as a newborn screening program manager might want to do these things, you have to have buy-in, and you have to have the funding available to be able to implement that change.

Also noted was the release of paper newborn screening results to submitters via the postal service. And limitations within the laboratory or the laboratory information management system functionality. And part of that is that ability to collect data to be able to measure these appropriately.

So, let's see. Some of the best practices, you hear --- many of these you hear over and over again. Expand operating hours of the newborn screening program, insure timely specimen collection and transit. Here's a new one, improve reporting and communications mechanisms, so electronic lab ordering, electronic lab reporting. Ordering is important because that cuts down on your

demographic entry time up front, so if you're not doing demographic entry in the lab that can't be a barrier to you getting results out. That may seem very simple, but it's not. It's a logistical issue.

Providing education to birthing facility staff on the importance of timely newborn screening. Providing cross training to newborn screening lab staff so that in case you have staff out in one area they can cover for each other in another.

Monitor performance, provide feedback. That could be the feedback we provide to ourselves as newborn screening programs, so not only to hospitals but to the programs, or within programs. And then, in general, newborn screening program improvement activities. And that's kind of that circle that was in the middle of that one chart. We need to look at all the processes.

So, following that we moved into the third part of the survey and looked at new

technology and second tier testing. So, has the use of new technology or adding new tests in your newborn screening program improved or hindered your ability to perform timely newborn screening? Nine states responded that it had improved their timeliness, 15 responded that it had hindered their timeliness. So the improvements noted, that a new instrument allows for continuous loading of test plates, automated instruments and assays that run any time during the day and overnight with minimal supervision, deployment of a new computer system, the ability to DNA results in tandem mass spectrometry for quicker results, and greater precision and accuracy which can lead to faster turnaround time. If you don't have to repeat tests as much, you can get results out faster.

However, here are the hindrances that were noted. So, the increase in the number of disorders increases testing time. So noted there is DNA testing for cystic fibrosis, so

that's a component of the test in most states at this point. And that is an add-on at the end, so if you do --- you complete your screening for IRT, then reflexes to DNA, that's another day or so of testing.

High cost of reagents, limited resources and capacities of newborn screening programs including staffing challenges, so I heard last night from a colleague that they've lost several staff in the last two or three years, and they haven't been able to replace any of them.

Pressures to reduce false positives leads to more testing before the release of results, and that's a delay that's observed. So that's, as I noted earlier about if we impact one side, we're probably going to have an impact somewhere else, and we have to really monitor that.

Second tier testing, again cystic fibrosis is noted here because cystic fibrosis second tier testing actually delays reporting

of the results. And CF at this time we don't think is considered a time-critical condition but we do have to get those results out as quickly as possible. And we'll be having that conversation very soon with the pulmonologists.

Okay. So, this note, our survey limitations. There was a lack of definition of terms, so when we went to the states and we said we want you to measure this, they said well, what does availability mean? What does availability of test results mean? You have 50 states, you have about 40 interpretations.

So, the data, as I mentioned in the Subcommittee meeting yesterday is not apples-to-apples. For most of those, you're probably not comparing apples-to-apples numbers, but it's a starting point for us to look at.

The lack of ability to collect appropriate data fields. So, that's something that as we move into our revised

recommendations that that will be mentioned. So, for example, in the laboratory if specimens are all received at the same time, you may not get a time of receipt, you only get a date of receipt. Well, that's going to limit your ability, so you automatically have to say day one versus eight hours.

And software limitations, so the inability of staff to quickly pull data for ad hoc requests such as this.

All right. So, we're going to turn it over right now and Dr. Dolan from March of Dimes is going to talk about their efforts before we move into this section.

DR. DOLAN: I appreciate the opportunity to just give a brief update on March of Dimes activities that are very much in keeping and in conjunction with the activities of the work group that we just heard about.

The first is the newborn screening quality improvement work group, which March of Dimes has been organizing. It's a quality

improvement work group dedicated to thinking about the culture of safety in newborn screening. It comprises 14 organizations, and I have the membership list if anyone is interested. So far the group has had two conference calls and an in-person meeting, and in addition there's a conference call planned for October. So, these activities are really in conjunction, and there's plenty of folks who are overlapping, and we thank you for your work on this Committee, and as well with the March of Dimes Quality Improvement Work Group.

As part of this promotion of a culture of safety in newborn screening, March of Dimes has initiated some awards that will be given out. And there's two general awards that the organization is going to be looking at. One are quality awards which are really policy awards given to state health officials when they have made the initiative within their state to set a target of 72 hours, 48 hours, or 24 hours for having the screening results

available, or having the policies in place to have the screening within those time frames.

So, what we're looking at doing is having a state be able to nominate when they make those efforts, and we could see a state going from a 72-hour award one year, and then the 48-hour award, and then the 24-hour award. The idea would be that it would be a progression, it would be the ability to set milestones, set targets, achieve them. And March of Dimes is delighted to be able to award and sort of recognize states that both have a commitment to full transparency, and then meeting those benchmarks.

The other award is going to be an annual award called the Robert Guthrie Newborn Screening Award, and it will be given to the state health official who meets the highest goal. And that actually will be given out at our Volunteership Leadership Conference which is meeting next week in Arizona. So, the first award will be given out. And that will not

--- that will be sort of an annual award recognizing an individual on behalf of their state. That's a different award than these other awards, where like I said, we would see states moving through the award process year after year.

This is a new initiative. We're still open to some feedback about it. We appreciate the input we've gotten from members of this Committee, as well as our work group, and we're delighted to be part of recognizing some of the efforts that states have put forward in response to the APHL survey results, this Committee's findings, the work group's findings, and then our Quality Improvement Work Group.

I was delighted to see yesterday that the timelines really line up, so we're not asking states to think of this Committee is telling us this, and March of Dimes is telling us this, which are we supposed to aspire to? We're delighted that, you know, the input has

been freely flowing between all these groups, and we're all on the same page in terms of what we're asking states to aspire to.

So, I'd just like to open for any questions about these initiatives, and thank you for the opportunity to present them to this group.

The process if anyone is interested is through Dr. Ed McCabe at this time who, as I said, sends his apologies for not being here in person, but due to a March of Dimes Board of Trustee meeting he's in White Plains, but he is very involved, as everybody knows. He and I work closely together on this, and for the moment the process for nomination is through Dr. McCabe or myself, of course. So, you know, feel free to contact us directly if we could speak with you about that. Thank you.

DR. KELM: Thank you. So, our last piece which may be the longest piece. So, the --- we've had lots of calls, sometimes calls have been an hour to discuss one recommendation

and how we might revise it. And then a lot of our time last night working with our group, our Subcommittee and some other people that joined us was also working on where we thought might be some ways to revision the recommendations based on some of the new data we have. But I think we also thought that there was still possibility of changes, because I still think there's some new data coming down the road that we've had a lot of discussions about that may change things.

We sort of had a eureka moment where we thought there actually should be a new approach. So, the emphasis should be on the goal of the program. So, we should actually move what we considered or called the four recommendations, three and four to the front. So, the goal of the program is timely notification of presumptive positives, as well as obviously completing all of the testing as quickly as possible.

So, whereas, Recommendations 1 and

2 are very important, these must be achieved in order to provide the means to achieve the overall goal of the lab, which is timely screening and getting those presumptive positive results to the physician as soon as possible. So, we decided to move them around, 3 and 4 go first.

So, the old one we have on the top, and we tried to pull out what we thought were some of the most important issues and things that we really need to deal with as we revise them. So, you know, what we kept hearing was this whole available, what's that definition? What do we really want? So, obviously, for recommendation 3 in order to achieve this, you know, it's all about collection, testing, reporting, all being timely. And we had this issue where timelines were too open for interpretation, so we needed to find these in order to down the road be able to capture this. So, what this recommendation really we felt should be is that presumptive positive results

for time-critical conditions should be reported to the child's healthcare provider within five days of life. Now, the only caveat is that, as we discussed, we're not sure that we can capture this data now, but we think that we can change --- you know, we're trying to change the system so that we can capture this.

Right now it sounds like in a lot of the systems this is free text that the physician was called and contacted, so if we can --- if people think this is more appropriate, which it's better defined, then hopefully we can then change the system so we can capture that.

It's a big one. All right, Recommendation 4. All results should be available within five days of collection. Once again, issue what is available? We had the same problems with definition, interpretation, et cetera. And as we discussed, we really felt that this actually had two components that needed to be separated out. So, it's important for providers to receive results on any out of range

or presumptive positive results for time-sensitive disorders in order to follow-up. So, we came up with this, you know, time-sensitive disorders indicates these conditions we screen for that aren't time-critical, but we felt this definition captured that timely screening for the disorders that also are important, all newborn screening needs to be done as quickly as possible. But it's also important for providers to receive normal results in as timely manner as they can.

So, we broke this one out into two, and I'm sorry this is sort of cut off on the bottom. All presumptive positive results for time-sensitive conditions should be reported to the healthcare provider within seven days of life, but also all newborn screening results should be reported within seven days of life. I think one of the notes that someone said was that when they're working with --- they saw research that was ongoing that said 50 percent

of the normals aren't always getting back into the patient's file. So, you know, that's still very important, and might not always being performed but should be captured.

So, Recommendation 1, initial newborn specimens should be collected at 24 to 48 hours of life. So, as we said, the considerations here, different recommendations exist for specimens collected from pre-term, low birth weight, and sick newborns, and we actually refer you to the CLSI guideline that exists for the best recommendations for taking specimens and treating those babies.

As we said, some states had different times frames in the regulations. We know that California actually collects at 12 hours, and after some up to 72 hours. And here's where we're talking about balancing false negatives and false positives, especially with endocrine disorders, but there was lots of discussion about getting data from California,

since they collect on the earlier side, looking to see how well they achieve, for example, detection of endocrine disorders with the fact that they tend to sample --- take specimens earlier than some other states. So, that might help moving things earlier, because the earlier that you can take a specimen, the faster you can obviously ship them and test.

The new wording that we worked on last night was that initial newborn screening specimens should be collected in the appropriate time frame for the baby's condition, but no later than 48 hours after birth.

So, Recommendation 2, specimens should be received at the lab within 24 hours of collection. Obviously, there are lots of limitations and considerations. We heard about the issues of couriers, geography, weather limiting this, as well as obviously the hospital staff batching and not drawing and shipping as quickly as they should. And there

is, you know, routine second screens, while they should be also received and shipped very quickly, obviously add a little hitch in here.

This one was the one that, as you said, we've seen the most issue with the data, and it has a lot of working pieces in order to achieve this goal, but it is still important in order to get the timely screening done to meet the first recommendation.

Our new wording here is newborn screening specimens should be received at a laboratory ideally within 24 hours of collection, but no later than 72 hours after collection. But what we want to look for here is that here's where states can make the most improvement in this recommendation which would, obviously, lead to better results for the overarching performance of the system.

So, here I've put them, the new ones that we've proposed all on a page for you to consider, discuss. And so we've put the new ones on the overall program up top, so in order to

achieve the best outcomes for babies, and we've put the reporting for the presumptive positives and overall results within --- you know, on the top. And that in order to achieve these goals and reduce delays we need to get collection and receipt ideally within those time frames.

So, we wanted to put these out for everyone to consider, think about, any discussions, we've love for your feedback.

CHAIR BOCCHINI: First of all, I want to thank Kellie and Susan for a remarkable bit of work within a short period of time. I think they have organized their working group quite well, and have been able to in remarkable survey results get involvement of every program which is, I think, very helpful to give us an understanding of the state of specimen collection, and that will be helpful to us. And, again, thank you for the work.

So, these are the recommendations of the Subcommittee for the Full Committee to now discuss. Our goal is to give feedback to the

Subcommittee, and see if we can finalize these recommendations, or ask for additional work to be done to complete them. Our goal if we're ready to do so would be to then have the final recommendations come forward to us at the next meeting for a vote for approval, and a decision as to --- let's open this to the Committee for discussion.

DR BOTKIN: So, a point of clarification. Do I understand that for state labs that the critical conditions tend to be reported out at the same time as other conditions, or do people --- or do systems report out results in a graded fashion depending on the urgency of the response?

DR. TANKSLEY: So, in general, there are --- so, there were --- trying to remember the number, 30 some odd states that actually have conditions they consider time-critical, so reporting is done differently. So, I'll give you an example. In Texas on Saturdays we process specimens, we do the testing, we get to a final

result. We don't report out a congenital hyperthyroidism on a Saturday, but we report out a whole list of other metabolic conditions, most of which are on the list recommended by SIMD along with CAH.

CHAIR BOCCHINI: Would you recommend that that be a uniform pattern, or is it already pretty uniform, or would that be a recommendation that ought to be considered, too?

DR. TANKSLEY: So, we didn't collection information on what states do with the critical conditions. We asked the question do you have conditions you consider time-critical? So, I don't know if we received any feedback. I'm looking at Kareema. I don't know if we received any feedback, you know, free text on that. I mean, the recommendation itself says that those should be reported out two days of life prior to the ones we would consider time-sensitive. So, I think that in itself tells you that you need to do something to be

able to report those out faster.

CHAIR BOCCHINI: First and then Charlie.

FEMALE PARTICIPANT: So, this is really an impressive body of work. Thank you all for putting this together. And I like the idea of emphasizing the most critical features first, which is really those time-critical conditions.

The only question I have that raises some concern is that the last two recommendations, if you add up the no later than 48 hours, and the no later than 72 hours, you're already at five days of age, so how can you possibly have time to do the tests and report out the results for a time-critical condition for those individuals? So, I worry that, you know, laboratories or public health departments literally interpreting those results --- those recommendations and not being as timely as would be ideal based on the first two recommendations.

DR. KELM: So, we had a lot of discussion about that. I mean, it's been brought up, obviously. You know, we had a lot of interesting feedback, and some of it is obviously --- we're hoping that all states will work to the lower end, as appropriate, you know, whether --- what you want to say. So, there's some states that you can improve that, and hopefully will do that. You have some states that won't or can't.

So, I think in also talking to, you know, NewSTEPs, the personnel that was there, I mean, obviously these are numbers that we can look at and report, and maybe use that for states to improve their performance. Find out what they're doing now and then use it to help them move in a positive direction.

It was interesting, I mean, they even showed that a lot of --- Recommendation 2 was the hardest one which is now --- we didn't number them but, you know, getting them to a lab within 24 hours. That was the one that they were

meeting the least, but of the ones that actually report time-critical conditions in five days, that number was greater than 80 percent.

So, although the math doesn't add up, I still think, you know, that's the idea in order to achieve these, you know, shrink these but, you know, we obviously also know about the issues with some of the states and getting things there in 24 hours.

CHAIR BOCCHINI: Charlie, then ---

DR. HOMER: I just first want to add my commendation, very exciting to see a survey with 50 responses.

I have a couple of small --- a couple of comments I'll group together. One is I told my Subcommittee that QI has lots of (inaudible) which drive people nuts, but one of them in response to your last observation about hope is we have a saying that says hope is not a plan. So, I mean, that sounds facetious but I do think in terms of the bottom two recommendations, hope is not a plan. So, I think

if we feel it's important my inclination would be that we stay the standard as --- come up with timelines or facilitate efforts.

On the first one, I guess, again I'm not a laboratory person, is presumptive, which is will there be universal understanding what presumptive positive is? Clarify that. The second two recommendations seem redundant to me. All need to go, all includes presumptive positive, so unless there may be a subtle distinction there.

DR. KELM: This is capturing, as we sort of mentioned, like CF needs actually the second tier testing, and a lot of those have second tier testing, so we would like to get them out even earlier than seven, but a lot of them require that the testing which still takes about seven days.

DR. HOMER: But, again, it says all presumptive within seven, and then it says all within seven, so the recommendation --- I mean, two is a subset of one.

DR. TANKSLEY:: There's a difference. So, the difference is that it is how you would calculate your results for that. So, it's different time points that are measured. So, if you look at presumptive positive results and reporting out presumptive positive results, it's reported to the healthcare provider. That means the healthcare provider --- you have made contact with the healthcare provider; whereas, if you look at all results, the time frame that you're measuring is to the time you have a report available. That report is not communicated in person to anyone, because those are your normal. So, that --- it's a subtle difference, you can drop one, but it's actually calculating --- it's calculating for your presumptive positives for everything else, and it's calculating all of your results, so you calculate for your critical results communication to healthcare provider, your time-sensitive results for reporting to the healthcare provider, and to

the time you have all results available in the form of a report.

DR. HOMER: So, then maybe it's just a question of wording. I think what you're saying is, basically, if you post to a website or send an email, or put a letter in the mail you would have met Number 3, and for Number 2 you actually needed to document the conversation.

DR. TANKSLEY: And that would all be captured in like the discussion, you know, how --- those things, like the definitions would be defined within the paper itself.

CHAIR BOCCHINI: Steve is next.

DR. MCDONOUGH: I'm indicating some disappointment in the recommendations. I don't think this goes anywhere far enough considering the problems that we have. Babies are born every day in every state. Doctors check babies every day, hospitals draw blood spots every day, babies have their hearing checked every day, they have their O2 stat checked every day. If

you're involved with healthcare babies you need to be open every day. Particularly for these time-sensitive critical conditions, state health labs need to be open every day, and there's no recommendation in there for that.

A baby's chance of survival should not depend on the day that they're born. And, unfortunately, in this country that's what happens, and we could do better than this.

I think that I would --- even though an example of just being in practice, a couple of years ago I was on call on a Saturday, and I received a phone call from the Iowa Public Health Lab which does North Dakota's testing, on a baby who was born in a small town near Bismarck. It wasn't born in Bismarck, on Thursday. And I got a call on Saturday afternoon on a presumptive positive. It wasn't even my baby, it wasn't even my partner's baby, it was in a totally different town, but they couldn't get a hold of the doctor there and they called me. And we had that child admitted to a

hospital, and this is within two days of age.

This is in a state, North Dakota, that doesn't even do public health testing for newborn screening. It's done in another state, in Iowa, so I know we can do better than the data indicates. And I think our recommendations need to be stronger.

I don't see why we can't have results of presumptive positives within four days rather than five. There should be a recommendation for public health labs to be open every day, and there's none here. And, unfortunately, the public health labs have been under a lot of stress lately. We had a great recession which put very strong financial pressure on the public health labs, and then they had to absorb the large workload of congenital heart disease screening, so I'm empathetic to public health labs of trying to improve what they're doing under very stressful circumstances.

I think it would be very nice if

federal funding sources or agencies such as MCH and CDC provide some financial assistance to state labs so they can do process improvements, improving either the number or opening --- being open every day of the week if they're not currently, or adopting new technology, or assistance in hiring staff.

Right now what we do in this Committee is we basically provide them kind of an unfunded federal mandate when we actually come to a conclusion and recommendation that actually gets approved by the Secretary, which doesn't occur on a regular basis. They are forced to try to implement that without us giving them any assistance. So, anyway, we have babies in this country who are dying unnecessarily because of the day of the week that they were born, and we should be doing better. And I think our recommendations should be ---

CHAIR BOCCHINI: Coleen.

DR. BOYLE: Yes, I want to recognize

the great work that you all did, as well. And I was wondering trying to --- being about data, and trying to help facilitate this process, have you considered as one of your recommendations actually having a tracking and management --- these recommendations are actually put into practice.

DR. KELM: I don't think we discussed that. I think it's something to consider. I mean, it might be worthwhile as, you know, something to put in the report, but I think for long term we thought, obviously, it sounds like that was a hurdle for now, but that, obviously, we were hoping that was something that we could fix in the near future. And that, obviously, the recommendations would be in place for longer than that, but we can have some discussions about where that --- we definitely think that needs to be available for states, figuring out the mechanism for recommending it, or at least, you know, putting it in the report. We can discuss that, but that's a good point.

DR SONTAG: Hopefully in response to that NewSTEPs is collecting this information, and as Susan mentioned earlier, we're working with the vendors to put this into their system, the quality indicators. And many of these stem directly from the quality indicators that we have opened to our repository, so those are things that will be reported out to the national repository on an annual basis.

It was also at the states' request, can we track some of these things locally on a monthly basis, and working with all of the vendors who are very interested in helping us with this, I think we can give them some tools so they can collect if we have it progressing monthly on some of these.

DR. BAILEY: Thanks for a great collection effort in getting this organization. In several of your slides you mentioned education and training as one --- and I'm trying to --- I'm wondering if you have any more detail on what that might be, because it

seems like the issues are much more structural and financial, and maybe --- awareness of the importance of working towards these kinds of recommendations, but I'm wondering if there are specific technical training kinds of --- talking about as well there, or --- anyway, I would just love any thoughts you might have about what that is, because I think Catherine and I both would --- Education and Training Committee to think about this at all.

DR. TANKSLEY: Thank you. So, much of the education and training that was mentioned was for the healthcare providers. So, for birthing facilities --- prior to that. So, you know, proper specimen collection, proper --- how do you handle it once you have it in your facility? What is the importance of having timely newborn screening so that it is not batched?

In Texas we learned --- so, we've always said don't batch, don't batch, don't batch, but there's different variations of

batching. There are satellite hospitals that feed into a main facility and the labs feed into that main lab, so you may lose a day or two just going from one hospital to the other. There may be systems in place within that hospital, because we've talked to a lot of those facilities and those facilities said well, how do we do that? How do we not --- how do we send directly to you, because they don't have the mechanism in place to track the stuff when it's within their own facility because they utilize the main facility for that.

So, as I said, there are different issues, and the more we dig, the more we find out. But a lot of it is education and awareness. The healthcare providers that --- these are the goals that need to be met. We're trying to insure timely collection.

We had another instance where we found out --- we provided feedback to a healthcare provider who had the over five-day delay, the huge percentage of over five-day

delay, and they said well, we thought we had 13 days to get it in to you. Well, you're missing the point here. So, I think there are issues with awareness of what is newborn screening, and why do you do it even within healthcare facilities. And there's a large staff turnover, so you can't educate once. You have to educate often, but programs are constrained.

We have over 500 birthing facilities in Texas. We can't visit them once, so how do you deal with those sorts of things? So, education and training of one sort for healthcare providers --- I don't know where Carla is at in the room. Oh, I'm going to let Carla speak for a second.

DR. CUTHBERT: Yes, thank you. A group of us were able to attend the AWHONN meeting earlier this year, and I forgot what AWHONN is. I think it's the Association for Women's --- it's a group of nurses --- yes, someone knows it. Thank you. Thank you, a wonderful, wonderful group, but I was actually

able to present with Emily Drake at a session for newborn screening. And we really just wanted to target the nurses, and remind them, and give them an opportunity to understand how to do collection of samples.

Prior to the meeting, we were able to get about 200 copies of the CLSI guidance document on blood collection, and we distributed them freely to the people who were in attendance. And we have a list of them that we're going to distribute the DVDs, as well.

As a follow-up to that, we secured a number of other copies, and we're looking at ways right now to strategize to approach the nursing community again to help them with quality improvement activities.

One of the nice things about this was that CLSI said that they would give permission to us if this was asked of them for the nurses to put the DVD onto their network, so some of the nurses were asking well, this is one DVD, that's great, but we have, you know,

multiple hospitals, we have lots of training opportunities. So, with permission they can actually put it onto their network and use it as a training resource for their entire program.

We're looking to find a way to encourage the nurses to distribute this in a way that we can actually capture quality improvement best practices among the nurses, and to be able to chart them. Now, we're just starting with the nurses, the birthing facilities would be a different thing. There are other conferences that address some of those other locations. This is just the beginning of the process for us.

MS. WICKLUND: This is just following up on that issue a little bit. You guys said some states reported back to you that they were already like educating the birthing facilities, and providing training. Did you get any idea how they were doing that? I know you said like in Texas it's huge, and you have tons

of places, and they're hard to visit. But I know that wasn't the purpose of your survey, but did you get any information about what is being done, and then how actually successful that was?

Careema Yusuf: As to how they're doing it, a lot of them online training manuals or they have annual meetings where they provide training to the birthing facilities, so they have booklets that they share, they have pamphlets to share, and things like that. But, again, the feedback that we received is that the resources to do that are reduced, so they're not able to do it as often as they would like.

CHAIR BOCCHINI: Can I ask you to identify yourself for the recording?

FEMALE PARTICIPANT: I'm sorry. This is Careema from APHL.

CHAIR BOCCHINI: Other questions from the Committee at the present time? If not, then --- oh, I'm sorry. Go ahead.

DR. HOMER Just a brief comment on

the education. Those of us who work --- and, again, I'm sure that's most people in the room in the behavior change field know that education is a valuable first step, but typically education is not sufficient to drive changes in behavior.

So, for example, I guess one analogy I'm thinking of is the work that a number of states have been doing working with their hospitals to improve the entry of electronic birth certificate data. So, again, part of it you start with education, why is this important? But then it really needs to move to feedback to actual data to the nurses at the front line to show them how it's important to give them the run charts, the data overtime looking at that, and giving them the power. And when we finally actually bring that data back to the front line providers who are doing that entry, you know, we've got places that are now turning around electronic birth certificate data much more --- I don't think education is

a key first step, but it's not the whole ball of wax.

DR. KELM: And I think that's --- you know, we are trying to work, and I know, you know, the group that March of Dimes convenes includes the American Hospital Association. We're still trying to reach out and maybe discuss with the Joint Commission about standards. And I'm sure that process is going to be a longer one than January, but there are some ways that maybe we can try to make it an actual standard, and something that's set that people are assessed against. But I think the feedback, obviously, there's some data being sent, but, obviously, the --- it sounds like some states want to do a better job. I don't know if they can, but more frequent data being shared. But, obviously, it was variable between the states as to how often they were doing that.

CHAIR BOCCHINI: Carol, and then Natasha.

DR. GREENE: First, I had kind of a

question for clarification to be sure that we all understand, then a specific comment about the draft new recommendations.

It is my impression from what you said that some of the states that did not reach their goal for having the sample collected between 24 and 48 hours, it was because the sample was collected early, and that's why you were hearing that one of the obstacles was that there's different recommendations for collection for the sick babies, which is to collect them the moment they hit the NICU, which could be an hour of age. Is that a correct assumption? If so, I think it just needs to be made a little bit more clear on how you report the data because the assumption that many people will make is that the states that did not meet that goal, it's because it was too late. And you want to be clear that some of the states are collecting --- don't make that goal because we've inappropriately said that you can't collect the sample before 24 hours of age, which

is against the recommendation.

DR. KELM: Yes, and I think we saw that that was a great number of them, but there is also some states that the regulations allow them to collect up to 72. So, some of that is that in some states it's acceptable to collect later than 48, so it's both sides.

So, there was some discussion about whether or not we would actually make this recommendation only for apparently healthy newborns between 24 and 48, but we felt this grasped the fact that depending on the newborn, it's appropriate for their condition, it would be --- it could be early in some cases.

DR. GREENE: Right. The recommendation I think absolutely captures that, it was your data collection slide where some --- you know, the percentage of labs that were not able to comply with the recommendation included a lot of labs that were getting samples from sick babies too early, and that should just be clear.

And then for the recommendations, speaking just --- it is --- I think the recommendations are great, speaking --- comparing them to what the SIMD statement is. I think it's possible to clarify a little bit, because I think it's not entirely clear the distinction between reporting a presumptive positive as soon as you get it, and no later than five days of age. It might be some language that would be appropriate. And, of course, for the SIMD statement, you know, it's going to vary on the laboratory. You know, if the C3 is 25, then you're going to, as most labs do, you're going to make a phone call and say I haven't even had a chance to rerun it to see if this is right, but I want to alert you, as opposed to a C3 of 8, and you're going to recheck it and make sure it's 8, and then call. So, each lab is going to have their internal processes as appropriate, but I wonder if you could incorporate the language that the presumptive positive report results for time-critical conditions should be

reported to the healthcare provider immediately on having a presumptive positive and no later than five days of life. And that will also help with the distinction between, you know, you might report a positive before you have the whole rest of the results, to maybe import some of that language.

MS.BONHOMME: Okay, thank you. This goes back to the conversation we were having around education, and I agree with everything that has been said in terms of the importance of reaching out to the nurses, and also the midwives who are on the front line of this, Genetic Alliance through Babies First Test. I've had an engagement with AWHONN, as Carla mentioned, the Association of Women's Health, Obstetric and Neonatal Nurses for over three years now in terms of presenting to nurses and working with them.

I think one thing that's important in the education is not just messaging out why is it important from the lab perspective, why

getting the samples back in is important, but also what the barriers, or assumptions, or the culture at the birthing facilities, because we have done focus groups with a birthing facility in D.C. that serves babies that are residents of D.C., Maryland, and Virginia. And one of the things that came up that would have never crossed my mind was oh, why does this have to be at 24 hours? Is this really just a way to keep the baby in the hospital longer? So, if you --- and that may seem oh, my gosh, to us, but if that is an actual belief at that ground level, as part of the education we have to find a way of addressing that. So, I think these recommendations make sense presented from the lab perspective, but there are probably another set of recommendations and strategies that could come from the education and training perspective that could really address the issue of what is happening at the ground level, what are the beliefs and assumptions and barriers there that we can then hopefully find some

strategies to address. So, I hope this Committee will take that up in some effort.

DR. BOTKIN: Yes, my thanks, too, for really very helpful and important work. It seems to me the recommendations are designed around outcome markers and goals, as opposed to process issues. And if I understood the data that you collected, there's a variety of challenges that these complicated systems face, but big ones were labs not being open on weekends, and courier systems.

So, sort of picking up on Stephen's comments, why or why not make a specific recommendation to say labs ought to be open on weekends, and that people ought to use overnight courier systems to achieve that? And I'm also interested, kind of noted in passing about the Joint Commission. It seems to me that's a pretty big stick. The hospitals really do care about that, arguably more than what they might care about from demerits from the Health Department, so I wonder if you have specific

thoughts on why those --- whether those might be recommendations to consider?

DR. SHONE: Excuse me, this is Scott Shone. So, I appreciate your comment, Dr. McDonough, about where the recommendations need to go, but I do think it's unfair to singularly point at the Public Health laboratory as needing to be open seven days a week, because you have scenarios where even in a scope of a laboratory that's open five or six days a week, you get the same outcome that you had with a three-day turnaround of a critical result. The fact that you can achieve that, you just don't achieve that 100 percent of the time.

But this whole discussion started out with a request for the work group to come up with what is the newborn screening system? And we need to acknowledge that this isn't a laboratory issue. It's not a program issue. It's a newborn screening system issue, which is what you were just addressing, Dr. Botkin. And

so in Texas you have 500 hospitals, and you probably have them do newborn screening collection 400 different ways. So, we could be open every day of the week, but if a hospital decides to collect on the first shift, and they draw those blood spots, and their courier picks up, you know, during the first shift, there's already a 24-hour delay. And you have a hospital that has timed it so that they collect it and send it out the same day that it's collected, but getting that message out to that part of the system, throwing it on the program, the laboratory, et cetera, negates that whole part of what's going on here. So, there has to be a multi-faceted approach to these recommendations that doesn't just target the lab, doesn't just target the follow-up program, but the whole system, the picture that Kellie and Susan put up in the beginning. So, while addressing one part solves one part, we're still going to have all the same issues, so in February when this group meets again, if all the

recommendations focus on having the labs open 24 hours a day, then we'll meet in 2016 and talk about well, how do we get the hospitals to collect in a timely fashion, and make that recommendation to look at their process to improve that. So, I think these recommendations are excellent, and I say that as being part of the group yesterday that helped come up with the recommendations, so that's full disclosure. But I think that it levels measurable and achievable goals to start at least from a program perspective, which is what --- but I think it needs to go wider.

CHAIR BOCCHINI: Do you want to address that further, Susan, and then Cate.

DR. TANKSLEY: I want to make a comment. These results, or these recommendations are achievable, but they're very difficult. These are not easy. If newborn screening programs set these as goals, we will move mountains. There are so many barriers out there that are difficult to overcome, and in the

last year a lot of work has been done to improve timeliness. And states have taken it upon themselves to do that. A lot of improvements have already been made.

When we collected the data, we didn't want to collect data from 2013. We wanted to collect data from the most recent, I think nine months is what we asked for, because we wanted more recent data that reflected the improvements that states have already made. So, if we took data from let's just say calendar year 2013, those numbers would have been much worse. And I know within our own state, we've made a huge --- a massive amount of effort in improving timeliness.

I think that setting these as goals for the programs will help all the babies in the U.S. And it may not be where we need to be, and it may be --- I mean, some of these conditions, we need a bedside test. And some of these babies won't be helped by newborn screening, and that has to be acknowledged.

There are some who, unfortunately, on the first day of life are having issues with some of these metabolic conditions, so I appreciate the suggested change in wording that Carol had that said okay, well, if you're meeting --- basically, what it means to me is if you're meeting five days of life, that's great, but that doesn't mean you should stop there. So, if you can do better, that's better. And I think we can use this as newborn screening programs to improve ourselves. We use them as our goal. Yes, we want all samples --- all specimens in the lab within 72 hours of collection, but we're shooting for within 24. And I think it's really important that we acknowledge that there are major barriers in areas. There are areas of states that have no couriers, so that within 24 hours is literally impossible.

We have received an email from a newborn screening program in Utah that said we have parts of the state where even the courier

takes two days. So, if you think about things like that, yes, it's easier when you're looking at urban areas. When you start throwing in rural, and you start throwing in hundreds of miles from the lab, it becomes more difficult. Yes, you can receive it if you have overnight courier available to that particular hospital, but overnight courier is not available to all. So, I don't think we should set programs up for failure that have even those barriers. There will be times that there are snowstorms where those couriers won't run.

We still have issues with couriers, major couriers, they don't run seven days a week, so if you don't have a courier that's there seven days a week, why have a lab sitting there doing testing seven days a week? We need to figure out how to remove some of those --- if we have any way to remove the barriers. There are barriers that you can't remove, so I just think that needs to be acknowledged. And that if states set these as goals, huge improvements

will be made.

MS Walsh Vockley: Thanks. Susan, I want to acknowledge the difficulties you've just relived for us again, but dovetailing on what Scott said, it seems to me that if we're going to work hard to improve all of the components in the newborn screening system, and certainly an acknowledgment of what parents have recognized as one link in the system that they have seen as being difficult in getting the samples to the laboratory, I'm still a little uncomfortable with codifying the 72 hours as the within 72 hours. I mean, is there data that you looked at from the materials that you collected that suggested that number? Could we not say 48 hours?

You know, is there some statistical way we could assess whether or not there's a better number to put there? It just makes me a little uncomfortable giving that outside time limit.

DR. TANKSLEY: Data were not

collected on any time frame other than the within 24 hours. It would be interesting if there --- I think states would kill us if we pulled another survey out of our pockets. So, it would be interesting to look at other time frames; 48 hours popped in my mind yesterday. Should it be something different?

Many states have that 72 hours. I mean, we've heard that, but where it came from was from that ACMG report, because there were two different recommendations. In one part of the report kind of buried within it said specimens should be received within 24 hours, or as soon as possible. And then when the actual recommendations were written out later in the report it said within 72 hours. So, that kind of gives that range of those two numbers.

DR. BAILEY: I think we're kind of in the middle of saying what's the ultimate goal that we want to achieve, and then how do we get there? And I would say that we should first

agree on the goal. There are lots of different ways to get there, and so to specify the, you know, the specimen has to be in the lab by a certain time, or the lab has to be open by a certain time; whereas, I agree those are all important --- it seems to me that the first thing we should --- the first --- what we really want to know is when the results get to the --- what we should be making, drawing kind of a line in the sand saying, you know, you can figure out how you can get there, give some examples of ways to get there, but here's the standard. Here's what we expect. So, I would focus on the first two or three bullets there, and not necessarily as much on the how to --- that could be a follow-up supporting document or something like that. But I think in terms of recommendations of the Committee, the most powerful thing we can do is to set the standard for --- the only question then is within --- is the five days, does that protect the vast majority of babies --- babies at risk

if we wait five days.

DR. TANKSLEY: There will always be babies at risk.

DR. BAILEY: Sure.

DR. TANKSLEY: But I --- Carol, I don't know if you care to make a statement?

DR. GREENE: So, first of all, I agree there will always be babies at risk. The conditions for which the time of diagnosis is most critical can --- all of them present anywhere from, you know, at birth, which is a little unusual but it certainly happens with some of the urea cycle disorders, to hours after birth, which is, you know, six, eight, twelve, twenty-four, thirty-six, seventy-two hours after birth for methylmalonic, most of the urea cycle, maple syrup urine disease, galxicimea. All babies with these conditions are --- with the classical presentation are typically sick around the time the sample gets to the laboratory. And what we teach is that the purpose of newborn screening is not to make the

diagnosis of the sick baby with MSUD, but so that when you're in the middle of your sepsis work up and you've already sent the amino acids, and the newborn screening laboratory calls you and says I think your baby has MSUD, you say ah, hah, but you've already started the treatment. Okay?

The newborn screening for these critical conditions is lifesaving for some situations, some of the babies with MCAD if the baby was perfectly healthy and you tell the family not to let the baby sleep long, the baby --- the unusual baby with galxicimea who was not already sick, and most especially CAH because they look fine, fine, fine, fine, fine, crash. Okay? But they're often --- they're typically already crashed in the NICU anyway.

So, the notion that the newborn screen is what saves the life of the typical baby with the classic form of these diseases is not actually correct. Those babies are typically sick by the time the sample gets to

the laboratory even if all of this is followed. Okay? That doesn't negate the fact that some of those babies are fine and will go crash a day later, two later, days later. Anybody who does what I do has saved the lives of babies with galxicimea by making a phone call and finding that the baby was home and getting a little sick. Okay?

So, we want to see --- SIMD wants to see these as quickly as possible, but we want to be really clear, this is not the major thing that is going to be lifesaving for the babies with the classic disorders. What's lifesaving for the babies with the classic disorders is family practice doctors, pediatricians, parents paying attention, calling when the baby is sick, and the clinician putting this on the differential diagnosis. This is all lagniappe, which is New Orleans for serendipity and it's nice it comes along with for finding the babies with PKU and all the disorders that present later with no symptoms. The babies with the

classic forms of these diseases are already sick by the time you do everything right.

I hope that answers the --- I mean, we still want the result as quickly as possible, but please nobody should be assuming this is going to save the life of the baby who dies on day one, or day three with MCAD or who is vomiting and comatose with OTC, or seizing with MSUD.

CHAIR BOCCHINI: This has been a really important and I think good discussion. And I think to sort of summarize, it sounds as if there's general consensus that the first three recommendations are appropriate with a little tweaking in language seem to be acceptable by the Committee, but that the last two recommendations, there's considerable concern about giving the margin of time out to the maximum rather than setting a standard that we want to achieve. I think that's the feedback that I think the Committee has made to the working group.

To answer Scott, I think that your comments are quite appropriate, and I think that we ought to be focusing on what we want, and let the states decide how they're going to achieve those goals. I think that's the most appropriate approach, but I think that what these recommendations do by allowing a delay in the maximum time that a specimen could be taken, and then gotten to the lab puts the lab in the worst position because then they have less time to achieve the primary goals that you have to get the results out in five or seven days. So, I think that we ought to be just as strong with setting a timeline for collection and receipt of the specimen, so that it gives the lab the appropriate amount of time to do --- that would be the feedback that I think ---

FEMALE PARTICIPANT: I might just suggest that, making a comment that this is a working living document, and that ---

CHAIR BOCCHINI: Okay. And, again, I want to thank you both for an excellent tackle

of this problem, and --- along very nicely. And I will also say that we are working closely with the March of Dimes, and with multiple other organizations to when we all come to consensus about how to move forward with recommendations, that's when the attempt will be made to go to the Joint Commission and --- requirements for the hospital side.

DR. BOYLE: I think maybe one last idea, and it's been mentioned --- our focus with this report has been more on the public --- the control of the public health laboratory. Couldn't we focus some attention on the hospital aspect of it? What we can do, performance measures --- bundled in. We did some work on Vitamin K, and the challenges with Vitamin K shots, and it's all bundled into one billing code. I just felt like we can do something ---

CHAIR BOCCHINI: I think that's a really good point. And I think based on the survey and the answers that you've gotten about

what the barriers are from the hospital side, getting to the lab, I think you have the genesis of the information that you need to kind of build on that. I agree, I think that's a --- it seems like that's a really component that has to be addressed. And I think that ---

DR. BOYLE: I would volunteer to lead a work group to think a little bit more about that.

CHAIR BOCCHINI: All right. I think that's a great ---

DR. BOYLE : We appreciate that. You know, a lot of --- I'm sorry. A lot of our --- you know, we were doing a lot of work and we were realizing that we needed to deliver something as soon as possible. Obviously, the scope was quite large if we wanted to handle, you know --- obviously, we have (inaudible) the public health labs and reaching at the hospitals was more difficult for us. And it was just a scope that concerning our day jobs, we were having trouble achieving that, as well.

So, I think that's still something that we are all interested --- obviously, very interested in, and we can talk about ways --- I mean, if the Committee wants to talk about that versus, you know, tasking it to us, but it was hard. I mean, obviously, most of our members are public health personnel and not so much with the resources of contacts in the hospital community.

CHAIR BOCCHINI: Coleen has just volunteered to get involved in that, so that's good. Okay, Carol, last comment, and then we need to move to the next item.

DR. GREENE: I would --- I've worked many, many times with our lab and our State Health Department, and our hospital, and education is key, but I'd like to come back to the JCAHO . You know, there are new people in pathology. If you have to re-educate them over and over, and they make the same mistake again two, three years later. And a question was asked about JCAHO yesterday, I think, from one of the

public comments. And I wonder if this Committee could possibly even vote on moving forward with working directly with JCAHO to make this a JCAHO sentinel event, that if samples do not leave the hospital in an appropriate time, recognizing all the other barriers of couriers, but if samples don't leave the hospital --- if the samples are not collected properly and leave the hospital in a timely fashion, if that were a JCAHO sentinel event, things would be a lot better.

CHAIR BOCCHINI: Carol, I appreciate that comment, and clearly that's one of the aspects we'd like to pursue. I think that what we'd like to do is really -- - we have a consortium of people through the March of Dimes, that includes the American Hospital Association, and a number of other groups, and so I think we would be in a much stronger position if that group together went to the Joint Commission. And I think that's what the general plan that's evolving will be, so I think

that --- time is appropriate. Certainly, I think we should ---

DR. GREENE: I was just thinking that if the Committee --- so, March of Dimes has huge standing and cachet, but if the Committee --- it might help the March of Dimes if the Committee were to make a statement that whatever the details would be would be worked out by that group, appropriately including everybody, but it might help the March of Dimes if the Committee said that newborn screening, getting newborn screening collected and out the door should --- failure to do that should be a sentinel event, and the details could be worked out by that consortium.

CHAIR BOCCHINI: Okay, thank you. Okay. Thank you, again, very much. Can you go to the microphone and give your name.

DR. MCDONALD: Susan is talking about Sunday deliveries of groceries and through the postal department, so I don't know why we couldn't just get, you know, you pack it

all up and it goes to one of these shipping things. It goes always the next day. I mean, it will cost 20 bucks per packet probably, or maybe 50, but ---

CHAIR BOCCHINI: I think you just found a new use for drones. All right, thank you. The next item is electronic standards for public health information exchange from the National Committee on Vital and Health Statistics. And I would like to introduce Dr. Walter Suarez.

Dr. Suarez is a physician and a Public Health and Medical Information Systems Specialist, and the Executive Director of the Health IT Strategy and Policy for Kaiser Permanente where he is responsible for coordinating and facilitating the development of Kaiser Permanente's internal and external Health IT-related policy positions, provide the U.S. National-International Policy input on Health IT-related domains. And fostering the establishment of and leading regional and

national public-private collaborative efforts on Health IT on behalf of Kaiser Permanente.

Dr. Suarez joined KP in 2009. Before joining Kaiser, he was President and CEO of the Institute for HIPAA/HIT Education and Research. And prior to this, he was CEO of the Midwest Center for HIPAA Education, and before that the Executive Director and CEO of Minnesota Health Data Institute. He also worked for the Minnesota Department of Health in various senior policy positions.

We've invited him here to present the background for a letter that this Committee has sent to the Secretary, and we felt after discussing with him that this is something that certainly it was important for us, as well, so we wanted him to make a presentation, following which I'd like the Committee to determine if we would be willing to write a letter to the Secretary in support of their letter. So, Dr. Suarez, thank you, and welcome to our Committee meeting.

DR. SUAREZ: Thank you. Thank you very, very much for inviting me. It's a great pleasure to be here. I represent the National Committee on Vital Health and Statistics, and Chair the Standards Subcommittee. And it's interesting the discussion that you were having earlier made me think of a lot of the discussions that we have at the National Committee, also. And, perhaps, the differences is in, of course, the domain. A lot of our discussion --- a lot of your discussions here today were about how to move faster and quicker, and reach the exchange of the samples, blood samples for testing.

We do a lot of discussion about how to make the movement of information, health information more efficient and more effective through the adoption of standards. And it's interesting that in the public health arena, the public health laboratories have really been a leading force in adopting standards for public health. And, certainly, the American

Public Health Laboratory Association, APHL, has really been the leading edge there.

So, what I've been asked to do, and I think I'm --- okay. So, what I've been asked to do is to present the recommendations that the National Committee made to the Secretary on Public Health information systems, and informatics, Public Health informatics standards.

I wanted to first do a brief introduction of the National Committee for those that are not familiar with the activities of the National Committee. Give a brief overview of public health information exchange standards, and where things are, in general. And then finish up with a review of the recommendations, and really appreciate the idea and the opportunity to talk to you about these recommendations, and continue to perhaps collaborate in the future in achieving those recommendations, and finding ways to operationalize them.

So, let me start with, again, a brief introduction of the National Committee. The National Committee is really one of the oldest statutory federal advisory committees in the country, was created almost 65 years ago as an advisor to the Secretary on primarily three areas, health data, statistics, and health information policy.

It has provided advice to several groups and organizations, agencies within the Department of Health and Human Services, the Data Council, CMS, CDC, AHRQ, and others, and it really has served and continues to serve as a forum where public and private sector organizations come to discuss and to present issues related to health data and information policies.

A few of the milestones really in 1949 was created in '74, the Public Health Services Act gave NCVHS, the official status of a statutory advisory committee. In 1996 with the signing of the HIPAA legislation, the

administrative simplification provisions of the HIPAA legislation gave NCVHS a very significant role in advising the Secretary in the adoption of national standards, and national standards across several areas. I'll talk about them in a minute.

In 2003, the MMA charged NCVHS with recommending electronic standards for ePrescribing, and then in 2010 the Affordable Care Act added some responsibilities to the National Committee on advising the Secretary on other areas of administrative simplification.

The Committee is formed by 18 members appointed by the Secretary for four-year periods or terms, and it's organized primarily in four core areas. The standards area which includes administrative standards for transactions, electronic exchange of information for administrative purposes. The codes that are used in those transactions identifiers, the identifiers to identify providers, patients, payers, others. And,

certainly, within those standards the area of Public Health, which is the one that we are going to be talking about here.

We also work at the National Committee on Population Health, and I'll mention a few of the items that have been developed under the Population Health Subcommittee. We have a Privacy, Confidentiality, and Security Subcommittee, and then we have also a Health Quality Subcommittee. In addition to that, the National Committee recently added a special work group advising the Secretary on all this new area around deliberation of information. You've probably heard about the Health Data Initiative of the Department of Health and Human Services, and the whole movement towards liberating information that federal agencies have, making it available. The National Committee created a work group on data access and use, and that's yet another area that we're working and advising the Secretary on.

We hold quarterly meetings, convene public hearings, listening sessions, workshops, and other roundtables, and other mechanisms to gather the information that we need in order to make an assessment and identify themes and issues, and then make recommendations to the Secretary.

Some of the recent notable activities, in the early 2000s, actually the National Committee was tasked to provide some visionary ideas about the direction of the Health Statics systems in the country, and we contributed to the 21st Century Vision for Health Statistics report published by NCHS. We also in that early 2000 had the --- well, HHS asked us to think about this whole concept of health information exchange, and the new notion that those early 2000 years of the development of a nationwide health information infrastructure, and health information network. And we actually made a number of recommendations that ultimately led in great

respect to the creation of the --- what's known today, of course, as the Office of the National Coordinator for Health Information Technology that really drives this whole area in the field of electronic health records, standards, and health information exchanges.

In the population health, one of the areas that we're working currently is communities as alerting health system framework, and we will be having actually a two-day workshop on October, I believe it's the 27th and 28th, on this topic, advancing really the community-level capabilities for using health information and becoming an alerting health system.

In the administrative simplification arena we've been working for over now 15 years providing oversight and advice on the adoption of the standards related to all these areas of administrative simplification. We provide to the Congress an annual HIPAA report informing Congress the

status of development and implementation, and achievement of simplification in the administrative arena.

Privacy and security, we have developed a national stewardship framework for health information privacy, sort of a high-level framework of the --- importance certainly of stewardship, and collecting, and maintaining, and using health information.

And this is a pictorial view, if you will, a diagram that shows some of the key elements of this vision that we had and provided as part of the development of the 21st Century Health Statistics report. Integrating really all the different domains and areas that we work with from community attributes to the contextual elements where the individual leads in the population operates, and the place and the time.

All right. Let me turn now to talk a little bit about the public health information exchange standards. So, as I was

saying, you know, one of the areas that we focus significantly on is really standards, but standards from the perspective of information exchange, and the movement of information. So, this is a collection of standards that support the electronic exchange of information between systems, between organizations.

And in the case of public health, it's certainly between public health agencies and the entity that provide and exchange data with public health. Nowadays, of course, we're talking more and more about multi, not just unidirectional exchanges, or submission of data, but bidirectional exchanges of information, and ultimately multidirectional exchanges of information to fulfill public health functions, core public health functions. And we're very pleased to see that a number of things have been now incorporated and adopted in the Meaningful Use program that requires health care organizations that are implementing electronic health records to

start to support public health information exchanges.

We're pleased to see that. We, of course, understand that public health is one of the areas and domains that health care organizations have to deal with in terms of collecting and exchanging information. And certainly, ultimately, the electronic health record primary purpose is to be able to deliver high-quality health care to individuals, and the exchange of information between electronic health records is made to support that type of quality health care services, and coordination of care, and transitions of care, and all these new concepts, some new concepts that really create a benefit of having exchange of information between systems that, you know, interoperate will improve that.

Certainly, with public health there is that goal, as well, of being able to exchange information directly from the electronic health record into public health systems, and

again receive back information.

There's certainly many public health data sources, medical data is one of them. The information collected on individuals at the point of care, the departmental data collected from many different sources, survey data you talked about, there's certainly a laboratory that all this different sources of information that is captured and collected by different entities, and that is of importance to public health.

The other aspect is the public health information infrastructure, and this is one area that we really focus on in our letter. I mean, certainly there's been a long history of the evolution, if you will, of the development of a public health information infrastructure going all the way back to the 1890 census, and there's a number of developments, of course, since then in the development of an infrastructure for public health, and for public health systems, for

public health agencies, et cetera.

What we have seen, certainly, and that is one of the findings and areas of recommendation is that we are still a long ways in achieving really a fully functional information infrastructure that truly interoperates. Not different from the clinical world where we also have that, and that is really the main focus of this major initiative that we embarked, you know, in 2010 with the High Tech Act and the Meaningful Use program. The significant investment that has been made in this country over $27 billion in insuring that electronic health records are adopted, use meaningfully, and used to exchange data has been incredible. Certainly, the challenge is the risk of creating by virtue of developing so rapidly this infrastructure in the clinical world, creating some technology, if you will, and ultimately digital divide, if you will, between that level of development in the electronic health record side and clinical

world with other very critical parts, like public health. So, that's one of the big concerns that we try to express in our letter.

Let's see. There's certainly databases and data integration, and this is part of the challenges that we certainly have heard and have been expressed in many different arenas, the state having separate systems, having developed separate systems for separate different purposes, from separate different funding sources. Many of those funding sources not necessarily, you know, long term sustainable, and so creating all this, you know, certainly silos and different levels of capabilities and developing, and support.

So, back in 2002, Chris Chute from Mayo, and Denise Koo wrote this statement that one of the serious shortcomings of these systems is the lack of horizontal integration, and the fact that ultimately data cannot be exchanged, linked, or merged across programs. And it seems like since 2002, over 12 years ago

now, the statement still seems to, you know, have some validity.

And what's interesting is in many ways this also again happens in the clinical world. And I think what we are seeing, certainly, is a lot more development in the clinical world to try to improve and avoid this type of inability or lack of easy, you know, integration of the data, but that also is a challenge, of course, in the public health field.

Now, you know, with respect to the systems, EHRs and administrative systems are sort of the --- so two, not all of them, but just two, and probably the two most significant sources of exchange of information with public health. And when we say electronic health record system, we're really talking about a larger scope of electronic health record system, not just the clinical system, but also the laboratory health information system, the pharmacy health information system, the

information systems used in imaging and radiology labs, and others. So, it's really much more comprehensive than what we have, you know, in the regulatory arena what an EHR means.

But in reality, you know, all these three areas interact and are there places where there's a lot more opportunity to insure an efficient exchange of information, particularly with public health, and again in a multidirectional mode.

Now, just getting a little more into the standard development side. You know, there's been a lot of activities, and there's actually a saying in the health care information standard world that the good news is we have a lot of standards. The bad news is we have a lot of standards, and you can choose from them. And then we have --- we used to have actually in the administrative world something that we thought was a standard for exchanging a basic element of business process, the health care claim.

We had a standard adopted under HIPAA back in 2003, and it was supposed to be the standard, and everybody agreed that that would be the standard, and we ended up having at least 4,000 different flavors, that's what we call, to that standard. So, every in this case exchange was between a provider and a payer exchanging claims. Every payer would say well, this is perfect standard. I see it, but I'm going to write a description of what I interpret to be what needs to go into the standard, because the standard had a lot of optionality, and situational elements, and ability, you know, to interpret. And when you standards that allow you to interpret things, well you have, you know, 10, or a thousand, or thousands of different interpretations, and that's what ended up happening.

In 2012, we moved to the next version of the standard, reduced the optionality, and interpretability, if you will, if you permit me to use that word, to by

at least 85 percent. And we are seeing now a lot more consistency and reducing the level of, again, interpretation and variability in the adoption of that standard.

Similarly, in the clinical world we're seeing the standard being adopted, and that have been adopted in Meaningful Use for the exchange of clinical information, not just with, you know, between clinicians but with other partners, that there is a lot of optionality, and it's creating again a barrier for interoperability, because I can have the best electronic health record system in my organization, but when it comes to generating a message that will go out, I generate it based on my own interpretation of the standard, and the entity receiving it will have to decode that in some way. So, it creates a challenge, a barrier, if you will, for that true full interoperability.

When we talk about standard, we talk about really standard that defines the message,

structure, the format, the quantum, the quoting, the vocabulary and terminology that goes inside, the transport mechanism, the electronic message transport mechanism, the security elements, and other elements. All those are the way in which we're going to exchange this information electronically, certainly so that the recipient can actually open it, and receive it, and process it.

Now, there's an interesting transition that we are all going through with the standards, too. You know, in the paper world you have a paper with the text in it, and someone has to read it, actually. Then you move to the fax system, and the fax system is nothing more than a faster way to move paper, basically, to get it from Point A to Point B. But at the other end, someone still has to see it, pick up the fax, and see it, and review it, and interpret.

The standards for electronic message allow you to include a print image, if you will, of that type of same text, or for

example a lab result. And when you send that message, you are not doing more than what a fax machine would do, which is sending a electronic message but inside that is not a structure message, it's what we call unstructure message. It's a print image, and so the recipient will still have to --- the system, even if you have the best electronic health record system will have to open it, and someone has to actually see it, and would not be processable in a automated way. What we are trying to move to is the ultimate level really of development which is 100 percent codified system, message in which every element in the message has a very well defined code. It's a structure message that can be received by the recipient and the system will open the message and execute it through electronic algorithms and the computer systems. And actually through mechanisms like clinical decision support be able to conduct some automated executable actions.

So, that's the ultimate goal. I

think we're still, as we all would say, this is really a journey more than a destination. We have to keep working on it, and keep trying to get to that level.

Apply to various health information exchange made between public health and external entities, and again moving from unidirectional to bidirectional, and ultimately multidirectional area.

Now, in public health certainly, you know, every instance where --- and we know public health just like clinical care is a very information-intensive sector, every instance where there's an exchange of information from public health and between public health and other entities, there is going to be an opportunity to use an electronic mechanism to exchange that information, and certainly a standard to do it.

So, here are some of the areas where there's been development, and certainly significant development in standards adoption,

vital statistics, someone was mentioning earlier the adoption of electronic birth record systems, and electronic death record systems. Interestingly enough, we are as a country not necessarily fully in that space yet, although we are moving quite fast to adopt those standards.

Immunization data certainly is one area where there's a lot of standards development, standards implementation, immunization registry systems across the country are using standardized messaging to allow providers to submit data to the registry about an immunization update or record, and then receive back, actually query and receive back immunization data.

Public health labs I mentioned. There are some areas where we're still very early in the process, particularly bio surveillance reporting, and public health case reporting. We really are at the early stages of development and implementation of some

electronic standards for the collection of public health case reporting.

Now, another area that is important to mention is --- it's not only about the exchange of the message, it's also about the ability for the provider to be alerted by the system that there is a case, if you will, or a situation that needs to be reported. And that type of clinical decisions before rules are an area that is I think a very significant area for work in the public health arena along with the clinical sector.

I think there is significant work being done in this area to try to get immunization registry systems, for example, to send back immunization and vaccination support rules, your clinical support rules, to the providers, so that the providers can actually be alerted about a particular vaccination requirement.

So there is a lot of, you know, closer interaction, you know, between

certainly clinical care and public health.

Now, a lot of organizations, just like there is a lot of standards, there is a lot of standards organizations that are developing those standards. On the left, you see most of the largest national and international standards development organizations, Health Level 7, HL7. Some of you might have heard of that organization; it's the one that develops most of the clinical types -- standards for exchange of information.

X12 is the one that developed the administrative standards for exchange of information. NCPDP, the pharmacy standards, an organization called IHE, Integrating the Healthcare Enterprise, developed the profile for a lot of the messaging that happens between providers that use these standards.

There is a lot of coding and code sets that have been developed. IHTSDO at the top developed something called SNOMED that is used in clinical care of course for codifying

the terminology system that allows the codification of a lot of the clinical text that we, as physicians and clinicians, type into our records.

ICD-10, of course, the national and international standards for federal classification, are its norm for the pharmacy area. The American Medical Association and the Dental Association have current provider or procedural terminology and dental terminology. So there is a lot of this standard.

The good thing is there has been a lot of conversions in the standards, and now basically there is a relatively limited number of those. And here is -- from the Office of the National Coordinator, this is the set of standards that have been adopted. At the top you can see, in terms of vocabulary standards, primarily these four are the core standard vocabularies and terminologies adopted -- SNOMED, LOINC, ICD-10, and RxNorm.

You can see from the content

structure there is a very, you know, limited number of standards being adopted for a specific purpose, and you can see lab standards being specifically noted as a standard for the content structure.

And then, there is the transport, the security, and other areas for ensuring the exchange of that information.

Okay. A lot of public health partner organizations have been working on the development of standards. And as I point in the next slide, I think some of the challenges that we see still are the level and the degree to which public health is able to participate in a lot of this development of standards. But, you know, all of these organizations have been very active.

A lot of them have joined and formed the one that is here in the center, JPHIT, the Joint Public Health Informatics Taskforce, which is an organization that primarily advises the industry and collects input from public

health in the area of standards for public health exchanges.

Some of the challenges for public health data standards adoption, I think that they need to move towards a more consistent standards-based data collection and reporting system from clinical systems to public health. I think a lot of the issues are really, in the clinical care arena, unless the data that is needed is for public health purposes, it's in the electronic health records system and it's included as part of the requirement of an electronic health records system, there is going to be difficulty in collecting that data and reporting it electronically. Of course, for public health you have to create specific collection systems.

The other part is really the internal workflows within the health care organizations that will need to be built in order to create the collection mechanism and the submission of the data to public health.

There is really limited local, state, and federal participation in standard development activities, although there is more coming, but this is one of the areas we wanted to highlight in our letter. Funding limitations to test standards that have been developed and to support EHR initiatives that include public health requirements is another challenge.

Really, even though it has been part of meaningful use, it is a relatively small component of meaningful use public health. And so so far we have three or four areas where public health is being included in meaningful use, but there is a lot of more expectations and need, really, to create exchanges between clinical care and public health, whether it's through meaningful use requirements or by virtue of basically creating the expectation that having an electronic health record system allows entities to exchange data with public health, all their data, not just the ones that

are mandated, if you will, under meaningful use.

Not all of the data certainly is -- that is needed in public health exists in a single data source or in an electronic health record. There is a lot of other data that has to be certainly collected. And some of it can be added to the electronic health record; some of it probably will not be part of an electronic health record. So that's an acknowledgement of a reality.

Not all of the data is also in electronic format. A lot of the data is on unstructured collection mechanisms and paper form still or printed forms not processable unless you have a data entry system.

And then, certainly this is something that requires long-time commitment. It is not a one-time quick-fix type of a thing.

All right. So let me finish up with the recommendations from the National Committee. So the National Committee held

hearings in 2013 focusing on the state of public health information systems and standards being used in public health. The hearings engage representatives from all the major public health professional organizations and associations and clinicians and others.

And it was really intended to -- in addition to understanding where things are and what things can be done, really create an awareness of the need to advance this whole concept of a public health information system across the nation.

So it provided us an overview of the state of affairs, a series of themes and observations, and in this letter that's what we highlighted.

Some of the themes were basically highlighting these five bullets here. First of all, the nationwide public health information infrastructure really needs significant attention and sustained investment. That was our key theme, if you

will. Even before we get into the -- what is the right standard for what is -- what method, we needed to highlight and step back and say, "We need, really, a very significant attention and sustained investment in the public health information infrastructure in the country."

The opportunity to identify and optimize common infrastructures, data analytic capabilities, and to avoid costly duplications was another area. We heard from professional associations, NACCHO and ACHL and ASTHO and others, that there is work being done to try to identify technology capabilities that can be shared across public health agencies, rather than each -- you know, each one of the 3,000-plus public health agencies having to reinvent technology or acquire technology having some common shared resources.

You know, and you all have heard, of course, about the cloud and about the type of services that are shared through cloud-based services. A lot of opportunities around that

coming to public health as well.

Need to establish a proper incentive for the adoption and implementation of public health standards, not just by public health agencies but by entities like provider groups and health plans need to implement a level of maturity and adoptability of standards for public health applications, and then they need to increase workforce informatics, competencies.

And so those were our themes. I'm going to be very brief around the recommendations. I know you have a copy of our letter, and I wanted to leave some time certainly for questions and for any ideas about how to move this forward, too.

Our main recommendations were around the -- first of all, this concept of having HHS develop and implement a new National Public Health Information Infrastructure Strategic Initiative. At some point, we compared this with we need a meaningful

use-type program for public health, not necessarily, you know, the kind of incentives and then penalty-type things, but some sort of a concerted effort to invest in public health information infrastructure. And that's what we made as our first recommendation.

Our second recommendation was around the creation of a dedicated fund, a Public Health Information Infrastructure Dedicated Fund. Again, at some point, we called it a Public Health Information Infrastructure Trust Fund, but the word "trust fund" seemed to create some political implications to it. So we changed the word "trust" to the Dedicated Fund.

But, clearly, the idea was to have HHS and to have the Secretary develop a strategy, a funding strategy that supports the first, you know, recommendation about a National Public Health Information Infrastructure Strategic Initiative.

So this fund will certainly support

the improvement of the information infrastructure capabilities across the nation, support public health information -- informatics standards adoption and use, look at where are the gaps in terms of the adoption of standards and the capability, information technology capability.

So the third recommendation -- well, we had actually a corollary to the recommendation, 2.1, leverage the Public Health Information Infrastructure Dedicated Trust Fund to provide continuous quality improvement for public health information systems, promote the development of sustained informatics skill, and then the standard development and adoption.

Our third recommendation was the creation or establishment of a National Public Health Informatics Standards Collaboration Initiative, again, bringing together all the parties into a collaboration initiative that will help really accelerate the adoption and

use of the standards in public health and will serve sort of as the focal point for leveraging the investments that will need to be done in support of public health information infrastructure.

So this collaborative initiative, we have some models and we actually have some organizations that have already been identified as possible places where this collaborative initiative could start.

Our fourth recommendation was about leveraging different policy programs and initiatives, such as the Affordable Care Act, meaningful use, to align the incentives for adopting and using public health standards and stimulating vendor engagement in the adoption of these standards, ensuring public health data requirements are incorporated into the standards, and certainly, you know, helping support public health involvement in the standards development and maintenance arena.

And then, the last recommendation

was about establishing a -- or developing a new national strategy for public health informatics capacity-building. Certainly, without the right trained workforce, all of this having infrastructure and having standards will not be sufficient. I mean, it will not happen. So we need a very well-trained informatics workforce in public health informatics, and so we thought it was very important to highlight this as a core recommendation.

So I'm going to stop here. Thank you, again, so much for the opportunity. I look forward to continuing our dialogue. Certainly, I have to say that I think there is a lot of area for cross-collaboration and continued dialogue and communication, and certainly the opportunity to have the recommendations be supported by an advisory committee like yours will be very, very valuable.

So thank you so much again.

CHAIR BOCCHINI: Dr. Suarez, thank you very much for your presentation. Clearly, the benefits of the goals that you have outlined for us are significant.

So questions, comments, from the committee or the (inaudible)? Steve?

DR. McDONOUGH: Thank you for your presentation. A couple of years ago this committee had recommended that, on a request I think from the Public Health Labs, that the national birth certificate include a field to link the newborn blood spot, and it would -- collaborative cooperation and information-sharing. And that was not approved by the Secretary. I think one of the reasons was is that the national birth certificate -- once every 10 years, and between 10 years it can't change.

Do you have insight if that is changing, if the birth certificate is so inflexible that it can't -- you know, will there be decades that it can actually change, or do

you have any knowledge about that?

DR. SUAREZ: Well, yes. I think it is one of the areas that the National Committee would want to come back and look into. We believe that there is the national birth certificate data set, and then there is the electronic standard that translates that. And we believe that there is an ability to really have the electronic standard, which is an expression, really, of data sets to allow for the inclusion of elements, such as the one that you mentioned, without creating any disruption in the structure of the national birth certificate itself.

So I think the -- that is, I think, where we are going to be looking at moving. I don't have any dates or any timeframes for when the next version or the next iteration of the national birth certificate will be issued, but it is certainly one of the areas that we will be looking at in -- basically in the next -- I think we have it in the schedule for the next

year at least.

PARTICIPANT: Thank you, again, Dr. Suarez. Any sense on -- or any response so far from the Secretary's office? And a related question would be sort of, how do you feel like this request is -- fits in with the work of the Office of National Coordinator? Are they supportive of the idea, or does it feel like a very separate thing from what they are used to doing?

DR. SUAREZ: Well, we have received work, really, that this has been very well received by the leadership of HHS. In preparing the letter, we had the transition of our Secretary. We actually had the letter written for the former Secretary Sebelius, and we had to change, actually, the header in the last minute, literally, because the letter was coming out a few days before or after the new Secretary Burwell was coming into office.

We have, again, heard from leadership at HHS that this was very, very well

received. A lot of the work that we do at the National Committee, and in fact the National Committee itself, is supported by the -- by ASPE, the HHS Assistant Secretary for Planning and Evaluation. And that is the arm of HHS that sort of leads some of these advances, if you will, in information infrastructure and information technology.

And so we heard from them that it has been very well received, that they are exploring how to operationalize the five recommendations, and they are actually looking for feedback and further I guess recommendations on how to specifically operationalize them. So from that perspective I think we are very well positioned with this Secretary.

With respect to the Office of National Coordination, we also had a transition of National Coordinators recently, and Karen DeSalvo, who joined the office a few months ago, she actually, as you all probably heard or know,

comes from the public health arena. She is the former Commissioner of Health in Louisiana and -- or Secretary of Health in Louisiana.

And so she actually -- we presented this about three weeks ago to her and highlighted the significance, and very, very supportive of this idea as well. So I don't think there is any doubt that this type of recommendations are going to elicit a lot more specific operational implementation ideas on how to move it forward. So I think we have, both from the leadership of HHS and ONC, a lot of support.

CHAIR BOCCHINI: Comments at the microphone, if you'd give your name and then the comment.

MR. OSTRANDER: This is Robert Ostrander, New York State Academy of Family Physicians, and I had a comment and a question. First, the comment is I would not put too much hope in meaningful use as a lever for this going forward, because you are going to see a huge

falloff in meaningful use participation.

We've had a great initial response to it, but the current next level standards are so difficult that a lot of -- there is a lot of pushback. A lot of practices can't and won't do it, and the audit process they put in place is incredibly punitive. And, basically, they take away your funding if you're not -- if you're 99.9 percent compliant, but not 100, they take away all of your funding. So I think you're going to see a huge falloff, and maybe you could also just advocate for a more user-friendly process, and putting whatever you want done in meaningful use through pilots first.

My question is, is there any thought of doing some of this information-sharing in a two-way approach? In primary care right now, we are seeing more and more -- within our practices we are looking at just what you all want, and what I think is a wonderful thing, which is public health and population

management.

And it would be tremendously helpful if there was a way to send some of the information back. You know, you tag it with the primary care doctor's name, and here is your list of patients with missing screening tests. Here is your list of patients who will be approaching -- due for the new -- for the next vaccine.

Here is a list of patients with this new recommendation that we have moved -- you know, "You've got to move (inaudible) down to 50 years old; this is your list of patients." It would allow us to do a lot better work with our patients. And if you could do this, it would -- that would incentivize us tremendously to want to participate in something like this.

Obviously, two -- establishing two-way streets, on the one hand it's a lot more work on the front end. I would hate to have it torpedo your efforts. But on the other hand, it probably would be more effective and easier

to do it at the front end than to do the sort of add-on an addition in a year or two when it's going to be clunky.

So any thoughts of setting up a two-way --

DR. SUAREZ: Yes.

MR. OSTRANDER: -- registry for public health exchange with -- actually, with boots-on-the-ground docs?

DR. SUAREZ: Thank you. Thank you for that comment and question. Very quickly on the comment, yes, I totally agree. I think meaningful use is one lever that might not actually be too much of a lever into the near future. And we included it as a recommendation just because it is still an important program certainly and will continue to be, but we do not believe that everything should be put in one single basket, if you will, in terms of that.

With respect to the population health management, absolutely, I think that is -- there is a great opportunity, and I think in

my view population health management is the next major transformational activity in the country. I think organizations, and speaking for Kaiser Permanente, organizations that have systems that collect information and have the ability to extract and do mining and data analytics in a way that allows population health management are going to be critically important in achieving, ultimately, the goals that we have in health. And the ability to transfer that type of capability to other organizations is also going to be critical.

So I think building registries and using registries to support that type of population health management and capabilities is one way I think it's going to be very important to both create the capabilities inside the EHR system, which is something that we at least have done, and others -- Mayo Clinic and others have done, and then creating the discipline to have the analytic capabilities to extract data, analyze, mine the data, and then,

like we do in our practices, every week, every morning basically, the system brings up specific population health needs of groups with respect to diabetes prevention or asthma or other things.

Those are the kind of things that I think the future EHR systems are going to have to have and build -- be built to support. So it's a combination of the EHR system and the internal systems being able to do mining and using data, and then interacting with registries externally, public health registries. I think that's going to be very good.

Yes?

DR. FINITZO: Hi. Terese Finitzo with OZ Systems. Walter and I have worked together for a lot of years. Sometimes it's hard for folks on this committee, including me, to see the forest for the trees, because this committee addresses life and death issues.

And so with that in our focus, don't

bother me with an information system and standards that don't appear to be really standard. So I think what we can do, however, is to perhaps decide what our biggest ping points are, and they should be those that hinder our capacity and capability to achieve best practices for our babies.

Screening is still overwhelmingly lab-based, but there is a lot that must happen at the point of care with these EHRs. I think what we can do is explore how to tie into existing public health standards and existing efforts, and I mentioned them yesterday -- the Office of Population Health, HRSA's OPH, CDC's National Center for Health Statistics, and certainly CDC's National Center for Birth Defects and Developmental Disabilities with the EDHI Program.

They are using some standards that might make sense to us. So I think what we could do is to take a look at how to leverage what has been done, so that we don't have to

spend a lot of time recreating things, and we can keep our focus on those babies. So that's what I would suggest as we move forward.

Thank you.

DR. SUAREZ: Thank you.

DR. ZUCKERMAN: I am Dr. Alan Zuckerman from Georgetown University. But I have addressed the -- this committee on several occasions both before and after our -- on these very issues, along with many people, including Clem McDonald, who is standing behind me, from National Library of Medicine.

At the -- and I think it's important for this committee to both join in with the recommendations and continue to work with other groups. And as part of that, we should remind the Secretary that the newborn screening formal use case was one of the initial activities in the NHII that over the years, in preparation for meaningful use, we did work to develop standards and vocabulary, but we have also learned how difficult it has been for newborn

screening programs and for hospitals who need to receive the data to adopt those standards.

We also worked to get newborn screening electronic quality measures into meaningful use. But another important role for this committee is to remind people that newborn screening is both a laboratory activity -- practices have caught up and are finally ready to begin communicating effectively with public health.

Thank you.

MR. McDONALD: I'm Clem McDonald from the National Library of Medicine and have actually been involved with this committee and lots of committees the last 30 years or so. But what I want to -- I want to reemphasize that this committee supported the standard for newborn screening, which uses conventional existing HL7 standards and conventional billing codes, and is being used and adopted by some states.

And if we keep talking about brand-new things, we'll never get anything

done. We've got to maybe push on with some of the things we've been talking about, too.

Regarding the public health, I am very involved in the standards organization, and I have -- maybe I shouldn't say all of this, but I have some opinions.

(Laughter)

And so public health is involved with the standard. I mean, they are -- not every department, but there is a fair amount of activity at HL7 from public health. In terms of meaningful use, there is more public health message requirement to public health than anything to the clinician. A clinician has just CDAs maybe, and so that you're not -- you know, the poor child in this whole process (inaudible) totally.

The second thing is, all we need is a fourth, fifth, and sixth standard organization, like you sort of alluded to. So I'd be very worried about if this is another czar, which just creates one more competing set

of standards. And I wish that thing was stated more to involve in the existing standard organizations and shape them towards your needs and goals.

And I fully support the need for more support, dollar support, because the biggest phenomenon we observed was public health wants these standards; they can't even adopt their own, because they are inadequate -- you know, they don't have the right money, they don't have the right funding, the right technology, the right people.

So I think the biggest -- biggest weakness is to make more standards for public health, but get some of the ones implemented with the appropriate funding and support for public health. That may not be a message people want to hear, but that's the way it looks to me.

CHAIR BOCCHINI: All right. Carol, last comment, and then we'll --

DR. GREENE: I can't say that I

understand the letter in detail, but there has been a lot of talk about linking newborn screening and birth certificates. And in the sense that that will allow us to make sure that every baby has been screened, or at least the screening is -- the screening is offered within the mandate of the state.

So I personally believe every baby should be screened, and that there is no good reason to object, but I recognize that some states allow for objection.

With that said, newborn screening and research around the data and making data public is an extremely sensitive issue for families, parents, and certain advocates. I don't see myself personally a distinction between the privacy of any information and the newborn screening, but it is a terrible red flag for many people, and attorneys general, and state health departments.

And I think we need to give quite a bit of attention to the issues of privacy,

confidentiality, and how families are involved before we start linking newborn screening with birth certificates. You know, even something as simple as the birth certificate is there, there is no newborn screen, and a phone call goes out to the family, we don't have a screen on your baby and it could be a name change, but it could be, "Why are you calling me? My baby died."

So I think there is a lot of attention that needs to be given to privacy, family issues, attorneys general, before we just move forward and link. That's not necessarily directly related to the letter and the goals of linking public health and EHRs, but at least in the arena of newborn screening there is a lot of sensitivity that should be taken into account before anybody moves forward.

DR. SUAREZ: Thank you. Thank you for that comment. We have our Privacy, Confidentiality, and Security Subcommittee looking into that level of issues of the

implications for public health data linkages, and I think that particular one is one that the subcommittee has looked into. So thank you for the comment.

CHAIR BOCCHINI: All of you have a copy of the letter written by the National Committee on Vital Health Statistics to the Secretary and have heard the presentation and comments.

And now that -- we can advise whether to send a letter of support for these recommendations to the Secretary citing the importance of these changes -- for the development -- our purview.

We don't need a formal vote. We just need a -- this would be to support the letter. It would be basically a collaboration.

Is there any downside? Is there -- I don't -- there is no downside at all. Steve?

DR. McDONOUGH: I move that we support the letter.

CHAIR BOCCHINI: All right. Second?

PARTICIPANT: Second.

CHAIR BOCCHINI: Okay. All in favor, just aye. All right.

DR. SUAREZ: Thank you very much, again, for inviting me.

CHAIR BOCCHINI: Thank you very much. This will end the morning session. We need to be back promptly at 1:00 p.m. to start the afternoon session. Thank you all very much.

A-F-T-E-R-N-O-O-N S-E-S-S-I-O-N

(time not provided)

CHAIR BOCCHINI: All right. Welcome to the afternoon session.

We are going to start, if we can get some slides up, with a brief review of Recommendation made by the committee. During our previous meeting, which led to a decision to have a vote, which, as you know, we had to schedule for a subsequent meeting, because it was not on the original agenda.

So, actually, while they're getting the slides up, let me do my roll call. Going beyond -- okay. Let's do roll call. Don Bailey?

DR. BAILEY: Here.

CHAIR BOCCHINI: Coleen Boyle?

DR. BOYLE: Here.

CHAIR BOCCHINI: Denise Dougherty?

DR.. DOUGHERTY: Here.

CHAIR BOCCHINI: Charlie Homer?

DR.. HOMER: Here.

CHAIR BOCCHINI: Kellie Kelm

DR. KELM: Here.

CHAIR BOCCHINI: Fred Lorey has been on the telephone, but has not been able to tell us that. Fred? And then, Joan Scott for Michael Lu?

MS. SCOTT: Here.

CHAIR BOCCHINI: Steve McDonough?

DR. McDONOUGH: Here.

CHAIR BOCCHINI: Dieter Matern?

DR.. MATERN: Here.

CHAIR BOCCHINI: Melissa Parisi?

DR.. PARISI: Here.

CHAIR BOCCHINI: Alexis Thompson? Cathy Wicklund?

MS. WICKLUND: Here.

CHAIR BOCCHINI: And Andrea Williams was also on the phone but could not let us know. Debi Sarkar is here.

MS. SARKAR: Here.

CHAIR BOCCHINI: Organizational representatives. Freddie Chen?

MR. CHEN: Here.

CHAIR BOCCHINI: Debbie Badawi?

DR. BADAWI: Here.

CHAIR BOCCHINI: (Inaudible.)

(Laughter)

Give me another three years and I'll get it right every time.

CHAIR BOCCHINI: Susan Tanksley?

DR. TANKSLEY: Here.

PARTICIPANT: Kenneth (inaudible).

PARTICIPANT: Here.

CHAIR BOCCHINI: Cate Walsh Vockley?

MS. VOCKLEY: Here.

CHAIR BOCCHINI: Carol Greene?

DR. GREENE: Here.

CHAIR BOCCHINI: Fine. Thank you all.

So at the last meeting when the final product of the subcommittee for -- Lab Standards and Procedures Subcommittee had

presented its final report on succinylacetone as primary marker to detect tyrosinemia Type 1, and the committee approved that report, but then the committee decided that they -- we wanted to send a recommendation to the Secretary regarding efforts to educate stakeholders on the benefits outlined by the report. This would be a way to try and make more people aware of the standard and promulgate it -- identify newborns with tyrosinemia Type 1.

So we have put together this recommendation for the committee to review, discuss, and then approve. The Secretary of Health and Human Services should facilitate a national dialogue among federal and state stakeholders on the benefits of measuring succinylacetone in dried blood spots to improve the specificity of newborn screening for tyrosinemia Type 1, conditioned on the recommended newborn screening panel.

DR. McDONOUGH: Mr. Chairman, I

move that the committee (inaudible).

CHAIR BOCCHINI: Okay. It has been moved by Dr. McDonough. Is there a second?

DR. THOMPSON: Second.

CHAIR BOCCHINI: Alexis Thompson. Is there any discussion?

PARTICIPANT: Is the definition of "stakeholders" obvious -- labs?

CHAIR BOCCHINI: Well, I think it would be the labs, but also be the newborn screening programs in each individual state based on how decisions are made in each state.

(Pause)

I think it is. Is anybody else concerned?

Okay. We will define the stakeholders in the letter to the Secretary. We will -- the recommendation, and then we'll identify the specific stakeholders.

Any additional discussion? Okay. So it has been moved and seconded, and now we

have to prepare for a formal vote.

Okay. So I know Don doesn't always like to be the first one alphabetically.

(Laughter)

MR. BAILEY: I'll save my --

CHAIR BOCCHINI: For more controversial issues? Okay.

All right. So before we vote, are there any members of the committee that have any conflicts of interest that would need to have them recuse themselves from the vote? If not, then vote yes or no to accept this recommendation. So Don Bailey?

DR. BAILEY: I vote yes.

CHAIR BOCCHINI: Jeff Botkin?

DR. BOTKIN: Yes.

CHAIR BOCCHINI: Coleen Boyle?

DR. BOYLE: Yes.

CHAIR BOCCHINI: Denise Dougherty?

DR. DOUGHERTY: Yes.

CHAIR BOCCHINI: (Inaudible) Klem?

Dr. KLEM: Yes.

CHAIR BOCCHINI: Charlie Homer?

DR. HOMER: Yes.

CHAIR BOCCHINI: And we'll see if we can get Fred's vote by (inaudible).

Joan Scott?

MS. SCOTT: Yes.

CHAIR BOCCHINI: Steven McDonough?

DR. McDONOUGH: Yes.

CHAIR BOCCHINI: Dieter Matern?

DR. MATERN: Given that there are so many different methods out there, so I think I --

CHAIR BOCCHINI: Melissa Parisi?

DR. PARISI: Yes.

CHAIR BOCCHINI: Alexis Thompson?

DR. THOMPSON: Yes.

CHAIR BOCCHINI: Cathy Wicklund?

MS. WICKLUND: Yes.

CHAIR BOCCHINI: All right. So the outcome is unanimous for those who are here.

Next on the agenda is the presentation by the Follow-up and Treatment

Subcommittee. Charlie? It's up to you whether you want to go up there, or we can move the slides if you want to sit there. You just tell Debi.

DR. HOMER: All right. So I'll try to be brief. We had a robust conversation yesterday at the committee meeting, and we've had a number of previous phone calls -- everybody who was on the committee -- their names quickly. I just wanted to review what our charge is as a -- just remind everyone what the charge is. That will become relevant.

Our committee was asked -- has been asked to engage in a multi-step process that identifies barriers to (inaudible) implementation, short- and long-term follow-up, develops recommendations to overcome those barriers, and offers guidance on the responsibility, who is responsible for what.

Our overarching charge in the (inaudible). We at the -- probably about two

years ago, there were a number of priorities that we explored. One was what's called Priority C, a look at the real-world impact and outcome of long-term follow-up, and we -- we chose to focus on exploring, to the extent which we can document improved outcomes, to determine whether in fact newborn screening is achieving the desired -- its intended purpose and -- evaluation of the impact of variability in clinical care. So that was our general concern.

That led to -- the next slide -- the substantial work which this committee has reviewed, which was the creation of a framework to assess the outcomes of newborn screening, whether we know whether we are achieving the (inaudible).

And that paper, which was reviewed and approved at this committee meeting, find what the key outcomes are building on previous work, previous papers the committee had done to find the key outcomes, which were survival and

well-being using the broadest definition of "health," including measures of disparity.

And we identified four critical drivers of those outcomes, including rapid and reliable diagnosis, which -- with evidence-based, with therapeutic and rehabilitative care, the coordination and integration of services, and then continuous improvement in knowledge generation.

So we wrote -- we put that -- we developed measures for each of those. We crafted measures -- reflect those specifically for sickle cell disease and PKU. The committee has -- that paper is pretty much ready for submission. We -- our team has been finalizing that. We're in the process of getting final approval.

So the question really is, given that we have now set up the framework, how do we move forward? What is our next step for that framework?

At the last committee meeting, we

discussed -- this committee approved the activity to be undertaken by our subcommittee, how -- further exploration of how to operationalize the framework. And the thought was that we would identify the extent to which there exists, both in public health systems and in clinical systems, and especially at the interface between public health and clinical systems, programs that are putting into place or have put in various elements of that framework; also, whether those programs not only have the measurement framework in place but are able to build -- to use that measurement to improve care based on the data that they're receiving.

So what we did yesterday was discuss -- on the next slide -- how we could go about doing that. And we came up with a number of critical activities, some of which are consistent with I think some of the other practices that we've heard about here over the last two days.

One was we felt very strongly that our committee needs to and should take advantage of the great work of the regional collaborative, particularly coordinating effectively with their Long-Term Follow-Up Committee, and also the close pulse that the regional collaborators have on the activities of the individual (inaudible).

We thought through that we could clarify which states have long-term follow-up systems in place, again, both to monitor and improve long-term follow-up. Through work with the regional collaboratives, we have identified barriers to more widespread implementation of such systems. And in discussion yesterday we identified a number of states which committee/subcommittee members felt likely had significant elements in place, such as Massachusetts, California, Michigan, Indiana, New York, and Rhode Island.

For those states that we thought had some elements in place, we wanted to identify

what capacity those states had to implement the framework, how they achieved what they did, whether -- were there generalized lessons that we might be able to apply elsewhere? So, again, that question of how the capacity could be extended elsewhere.

Then, we had a lot of rich conversation that raised a number of points that we're going to have to wrestle with. One was, are we focusing on what already is in place? How do we balance focusing on what is already in place versus some of the enormous potential for some of the things that are being put in place? That tied to the conversation, again, that was held in the full committee, how do we connect to and accelerate the adoption of the LPDR work that was discussed yesterday. Thinking about clarifying over the next (inaudible).

And then, just to highlight that there were several additional concerns that kept -- that have kept coming up in our

committee's conversations that I wanted to at least share with the broader committee, either for your feedback or guidance or simply to let you know these are things that are on our table.

One was, again, the framework that that paper articulates is about, how do you use measurement to drive improvement, that there is ongoing concern expressed by significant members of the committee that we not exclusively focus on measurement, but remember the purpose of measurement is for improvements and better outcomes.

And one reason that I wanted to start today's presentation with a reminder of what our committee is tasked with, we have -- also, a number of strong voices on the committee have highlighted that the broader charge of our committee is to identify and address barriers to long-term treatment, including the supply of and access to appropriate care and expertise, and that that not be -- whether that will be a separate activity, either to follow on or in

parallel, or whether we have a way of combining that with the current effort, is something that we need to --

So why don't I open it up, first, to members of the subcommittee and see -- oh, we should identify a workgroup that was going to work specifically on this idea of identifying promising states that framework they have in place. And these are handwriting -- the list of those who were on that workgroup, but there may be others that -- there may be others that I nominated that didn't know they were nominated. There may be others that -- so we will -- we can revise that.

But I do want to, I guess, throw this open to the members of the subcommittee first to clarify either errors of commission or omission that I may have had in the -- Alan?

DR. ZUCKERMAN: Alan Zuckerman from Georgetown University. I did want to add that we did spend some time looking at potential connections for the NCVHS letter

on standards and the fact that it might provide funding for implementation of standards and funding of (inaudible), and, in particular, the frustration that states have getting the funding to get data entry and to capture data entry once rather than to have duplicate and triplicate entry of data for long-term follow-up.

So hopefully we will continue to look for ways to interface the REDCap database that maintains the LPDR with the existing EHR systems to decrease the working cost of getting more complete funding. But we also tried to focus that the framework is not about the methods of data collection, but asking the right questions, identifying what data elements are worth collecting if we are going to do this without sufficient funding to get long-term follow-up at the point of care.

DR. GREENE: A lovely synopsis, and of course for those couple of slides at the end about what we want to not lose sight of. And

I just wanted to point out that -- implementation of the framework, the framework actually isn't published yet, so we can't really check and see how states have implemented it.

And so I think it perhaps might -- it's very nuanced, but perhaps more might be -- that with the framework in mind to look at what states are doing, there was some discussion about, you know, could states -- some of those states might be willing to look at the frameworks. So I don't think we want to ask ourselves whether states have implemented something that isn't published yet, but with the framework in mind as a stepping off point, what are states doing?

CHAIR BOCCHINI: Questions or comments? Let's open this up to everyone. Coleen?

DR. BOYLE: Yes. I have a couple of thoughts on the framework. And it's too bad everybody doesn't have it in front of them, but,

you know, basically, it's the potential measures to -- four drivers for long-term follow-up to essentially evaluate, you know, how -- how well we are -- and I was thinking back, yes, this is (inaudible) two conditions, sickle cell and PKU.

One thought I had was we have these great (inaudible). You know, perhaps -- and not that this committee would do this, but maybe the key -- might want to recommend that frameworks be developed for the other (inaudible) newborn screenings (inaudible) metrics for all of the conditions. So that's one idea.

And then, another idea that I was thinking was that maybe, again, just taking sickle cell disease, I want to think about the next steps here. Whatever our levers are to try to institutionalize -- two or three key performance measures across the life span -- maybe not the life span, but perhaps the childhood life span of an individual -- think

about. And I know there are performance measures.

Whatever our levers are that try to, you know, make these -- help influence the care of children with sickle cell. Maybe that's a next step for this. I think it's great work, but I was just thinking that it would be nice to have -- through it and perhaps do it in two different (inaudible).

CHAIR BOCCHINI: Thank you. Marci?

DR. SONTAG: Marci Sontag from NewSTEPS. I have -- for the long-term follow-up piece, I wanted to remind the committee of Beth Tarini's (work, which she presented to this committee a year ago, on long-term follow-up. She did a survey of the states. It shows a lot of great data that are really going to help inform the work that you are doing.

So I -- Charlie, I will forward you the slides that I just found from last year's meeting, and then you can use that before you

go out to the --

DR. THOMPSON: And I -- I think it was a very vigorous conversation yesterday. You know, but I must say, you know, especially in the context of (inaudible) sickle cell as an example, you know, it gave me pause, because I think that on the one hand, yes, in principle, the framework was meant to be developed and then tested to determine whether or not it is appropriate. And I don't think that we've actually determined that at this point.

So the investment in time and effort to look at it in other diseases, I wonder whether or not we should take a few steps back on that one, because again, you know, there are lots of good reasons why the two examples that were chosen will be very informative.

My concerns are that they are likely to show that we are nowhere to actually long-term follow-up, and that we need to sort of just acknowledge that that is what we are likely to find is -- that is, that we don't do

a good job of long-term follow-up.

And so finding out what the answer is for one or two conditions is likely to help us. Before we venture down the path of saying, "States, you should do this," I guess when we started this it was not my impression that we were looking at creating a framework and telling states what they should do, by no means.

It was really just to understand, be able to structure an environmental stance so that we could really understand, really, where our weaknesses are and then to look at what the opportunities are across a very wide system or in fact providing care for those (inaudible).

CHAIR BOCCHINI: And I think that's a good point to bring back to the subcommittee, to really decide -- to look at the effectiveness of this, have something in place, fit this framework in that to -- Carol?

DR. GREENE: Along the same lines, the frame -- I'm pretty happy that the framework is likely to work, but we never actually even

-- I mean, when you read the framework that was approved by the committee and is going to publication, we didn't set any specific measures or goals. So we said, "As an example, a measure might be X number of babies diagnosed by X day." And we didn't fill in the number, and we didn't fill in the days, and that will be disease-specific. It could be state-specific.

So, and I think Charlie has in mind that some of the things that we could do -- you know, we have to give a synoptic summary. But whatever the project might look like could involve having states proceed as this operationalizes. And I certainly -- personally, I agree we are not ready to try to develop it in specific for other diseases. We want to test the framework.

Personally, I also want to be sure that as we are moving forward on the framework we don't forget some of the -- you know, as we've moving forward on improving the data

collection, we don't forget that there is other ways to work on access issues.

But, yeah, I think what Charlie described has in mind what Alexis was just talking about, that the framework is a big picture, and we have to see, does it help states understand what is going on.

CHAIR BOCCHINI: Questions? Comments?

DR. GREENE: I haven't thought this through completely, but seeing that with the states makes me think that for some reason -- and I can't articulate it -- it might be good to include some states that might not be (inaudible) groups for quality improvement --

CHAIR BOCCHINI: Very helpful. Thank you.

All right. Charlie, thank you very much.

The next committee report is that of the Education and Training Subcommittee, and Cathy Wicklund will provide that.

MS. WICKLUND: Thank you. I, first, want to acknowledge all of the hard work that Don Bailey had done on this committee before I took over, and for giving me this wonderful opportunity to lead the Education and Training Committee. Thank you, Don. You're a giver. You're a giver.

(Laughter)

So Beth and I -- I am the chair of the -- the new chair of this committee, and Beth Tarini has agreed, with blood, that she is staying on as co-chair of the committee.

And so we had a pretty short agenda -- next slide -- in the sense that we are completing several of our priorities. So it was really kind of, you know, providing updates from individuals but also just kind of looking at the final steps of the two priorities that we have, and the one remaining, and then spending some time thinking about things that we can kind of tackle in the future.

So that's where our committee is at

right now. Next slide?

So we have three different priorities, and if you guys remember, Priority A was to track, provide input on, and facilitate integration of national education and training in initiatives, and there is further explanation of this.

And what -- actually, one of the -- next slide -- one of the initiatives that fell underneath this priority was the initiative that Beth presented at the last meeting, which was to identify childhood conditions that could be screened -- I guess the question was, could these be screened for during childhood. And if you guys recall, she did a nice summary looking at -- we looked at Fragile X Syndrome, Wilson's, and along Long QT. And she gave a summary about those conditions and some of the findings that we had.

So I'm not going to go through all of that again, but basically the charge was to identify heritable conditions that are not part

of the RUSP for which screening and treatment most likely would occur at a later point in child development.

And this is also getting at the issue that it's not just about newborn screening, but it's about heritable disorders in children as well.

The three conditions were chosen to represent a variety of clinical characteristics, including age of preservation, age of diagnosis and clinical morbidity, and so we went ahead and looked at those conditions for which Beth reported back on and -- next slide? And these were the six questions that we asked for each condition. And I'm not going to read through those again. We've talked about them several times. Next slide?

So after we presented this information to the committee at the last meeting, there was a request that we frame it in a way in which highlights some of the

barriers of doing population-based screening for these childhood conditions at this time, because the end result was that we really are not in a position to do population-based screening, and also looking kind of at like, what is the role in -- of public health versus the role of just having practice guidelines in general.

And so Beth put together a two-page summary of the findings, and that was given to us yesterday at the committee meeting. So the committee -- the subcommittee needs to kind of further look at that and just make any edits or modifications to that document, and then we will be submitting that to the overall committee to take a look at that. So that's ongoing.

And we also had a discussion, then, of what really to do with this work. So, you know, we have this work that has been completed, and is there something else that we should be doing with it. So Don and Beth are going to

discuss the possibility of writing a white paper summarizing the work of this initiative, and really discussing the role of public health in childhood screening versus the role of practice-based guidelines.

So that's going to be the next step, and they are going to come back to us with what they kind have decided about next steps in that process.

Don, is there anything you wanted to add to that?

DR. BAILEY: I think our goal would be, you know, an article that I think we -- these three conditions are very interesting, and what we did I think will be useful and interesting to the field. I think what we haven't done is take each one of them and think very specifically about (inaudible) of each in terms of next steps or recommendations. And so this would force us to do that, and then come back to the committee with some (inaudible).

PARTICIPANT: So this is very nice.

I'm just thinking from the health care delivery perspective, and quality improvement, and all of that, that the concept of practice guidelines is a little narrow, because a lot of other things (inaudible) evidence-based preventive services is going on or should be going on (inaudible) organization.

So practice guidelines is kind of a way to get things going -- done from about or 10 or 15 or 20 years ago and broaden that context. You're talking about the health care delivery.

MS. WICKLUND: Right. And take that with a grain of salt in writing down, but I think Don and Beth are going to look more broadly at that issue a little bit. But I think that's an excellent point.

All right. Next slide. So Priority B was really to promote newborn screening awareness among public and professionals. Complete. We're done, guys.

(Laughter)

Everyone is aware. Our job here is done. No, and I -- but let me say that the initiative that fell under this priority is complete. The priority itself will never be complete, but the initiative itself, and that was to support some of the ongoing efforts through the CDC and different things that we have completed.

So that -- we don't have any other initiatives underneath that particular priority at this time. Next slide?

And then the last priority was to provide better guidance for advocacy groups and others regarding the nomination and review process. Next slide?

So there are two things that are going on underneath this initiative, and one was the public-friendly document of the advisory committee's process for nominations. And that was something that was also discussed in collaboration with the Condition Review Group.

And so we have not started working specifically on this initiative yet, because we also want to recognize that, like Natasha with the Genetic Alliance, and the Clearinghouse is also working on this initiative. So, really, at this point in time, we want -- and the Clearinghouse just got awarded, like the other day, Clearinghouse grants.

And so I -- our next steps really are kind of wait to see what Genetic Alliance -- and have further conversations with them, because we certainly don't want to be repetitive in the work that is being done by the Genetic Alliance, and see how we can kind of support them in this process. And, again, this remember was to really give our advocates better guidance about how to nominate conditions, what the committee is looking for, what are frequently asked questions about this. So that's where we're at right now.

And, Natasha, did you want to add anything to that statement?

MS. BONHOMME: No. I think that it's just the timing of everything that -- and in a couple months I'll have more conversation (inaudible) and also with the committee -- exactly where should the overlap be in this --

MS. WICKLUND: Great. Thank you. Next slide?

So the other thing that is underneath that initiative is to develop a glossary of terms to be incorporated into the website. And this is to help, again, people -- there is a lot of terminology in there that might be above the reading level that we are really going for.

So Jeremy has spearheaded this effort thus far, and the reading level that we are really trying to go at is a sixth grade reading level. So we are trying to basically look at the terminology that is being utilized, like in the nomination form or different things, and then link some of those terms with like a definition that they can perhaps --

advocacy groups can look at and get a better understanding of what we're asking for in particular.

So Jeremy has a first -- actually, a first or second draft that has been going around, and Cate has volunteered to help Jeremy with some of those definitions. And part of the thing is, of course, you can't build on definition and definition and definition. You can a little bit, but each definition from a reading level needs to kind of standalone.

So if you put it through some of the different assessments, you know it's a difficult thing to get this down to, obviously, a sixth grade reading level. But they are continuing to work on that.

We did have a little bit of a lag time with that because we were unsure if we could actually do this on the advisory committee website. So there are logistics that we just need to figure out. When we are thinking about incorporating these, can we put

them on the advisory committee website, or is that going to have to be something that goes on the Clearinghouse?

So there are some implementation kind of logistical issues that we need to be cognizant of as we move forward on these initiatives. Next slide?

So given that, our -- you know, many of our initiatives have been completed. One of the things that Beth and I talked about was to really start a discussion on the -- what kind of are the current trends and barriers in newborn screening to help us identify what might be the next steps within the Education and Training Subcommittee. Next slide?

And what we did yesterday was just a really preliminary need assessment. We had put together about five or six questions that, if you guys had done strategic planning, recognized these from your basic strategic planning, really looking at issues facing newborn screening today. You know, what are

the most important issues facing education and training, and we left that fairly broad. That could be education of public, education of health care providers and different stakeholders. And, you know, where is newborn screening today. And this was really, you know, obstacles pacing us, kind of just to get us beginning discussions about what are the issues.

And this is not that we have decided these are the issues that we actually need to address yet. I think this is kind of getting the issues out there to help us think about, what do we want to try to tackle, where can we make the most difference, and those sorts of things.

So we had about a -- I would say a 30- to 40-minute discussion yesterday, just from the committee, and what I did was try to distill some basic themes that came out of our discussion. So the next slides are just some of the issues that kind of came out of those

discussions.

So, you know, one of the questions we asked ourselves was, where do the most serious issues happen in the actual system itself? And, you know, the fact that we are dealing with rare conditions, and of course the challenges of educating stakeholders -- and that came up a lot, you know, the knowledge of the primary care provider of the rare condition, being able to recognize these conditions, and this speaks to the timeliness issue that we were talking about before.

The education about and the impact of false positives are inherent, so that balance between, you know, giving out results in a quicker fashion but also about the impact of false positives, and recognizing that there has been work in these areas as well.

Okay. That's one thing I want everyone to realize. It wasn't that we thought these have not been tackled by other people, that other organizations have looked at these

issues, have gotten some initial data. And before we decide to do anything, we really want to be cognizant of that and to be able to get all of the data that has already been generated before we move forward on anything.

There was a lot of infrastructure issues that came up, and just also issues about state health departments, them being overwhelmed, lack of funding, some vulnerability, in particular with recent, you know, dry blood storage (inaudible) issues, and what the role of the media plays and the messaging that the public is getting about when something happens that is negative.

You know, you hear so much about that. You don't certainly hear as much about the positive things that state departments are doing, the IT needs that they have. And, again, just in general, all of the issues that go along with the current infrastructure that we have.

And workforce came up as well, and

I see that it came up with the long-term follow-up as well, so I'll talk about that in a second.

The other thing is education in general. You know, we talked a lot about the issues of education, and, Charlie, you said earlier, too, you know, just education is not enough. And being really aware of, what does that mean, and what are the other things that need to be in place, and how do we measure outcome and success. And obviously there are many, many organizations that are looking at this issue, and being aware of that.

What are the key relevant messages? How do we utilize parents and public and storytelling and getting -- you know, utilizing and galvanizing that population? And a lot of people listen -- will listen a lot more to someone telling a story than us as professionals telling them that this is what we think is going on. Next slide?

Again, the primary care provider,

access to the specialists. And, again, this all gets back to like the just-in-time point of care that we need to be aware of. Are there other models that we can use, you know, about other critical issues, for instance, infectious disease, HIV, and how, you know, hotlines and different things that we could be thinking about.

The timeliness issues obviously came up. You know, we had read the report that the laboratory committee had done, and so there were a lot of bullet points within that report that referred to education and training, and so that's on our radar is how can we contribute to some of the recommendations of the burning facility issues that came up.

And, again, I think the real thing is, like, how -- what impact can we have and how can we help there?

And then, the other thing that came up was the genetic workforce issues in general with regard to access to medical geneticists

and the growth of that profession, and then also genetic counselors, and we talked a little bit about the efforts through NSGC and the program directors group for genetic counselors and what has been going on with regard to looking at workforce.

And I know that ACMG convened the Banbury Conference recently, I believe like in February or March of this year, looking at issues regarding that as well. So is there anything we can do to contribute to that?

And then also, in general, just education regarding the exome and genome sequencing. So, you know, there was concern that sequencing -- obviously, there is more sequencing being done. We have the newborn screening sequencing grants that are out there. We have seizure grants, eMERGE grants, they are all looking at sequencing and integration to electronic health records, and also decision-making about return of results and what needs to be returned.

And not just education from the public and patient perspective, or consumer perspective, but also provider perspective and individuals who are ordering these tests that might not be as knowledgeable in genetics and really the interpretation and what to do with the results. So, and do we have a role in looking at that issue as well?

Next slide?

So, again, I just want to be -- we really I think want to be mindful of, you know, not doing work just to do work for work's sake, right? And really being mindful of, where can we have the most impact or influence in this area, and really continue to be aware of other organizations' efforts and not be repetitive in our efforts.

So is it our job to be a catalyst? Is it our job to bring people together? You know, can we leverage, you know, the different organizations that are represented in this group? And how can we maybe, you know, find

synergy there and, you know, get people to work together towards some of these goals? And, again, not losing sight of, where do the heritable disorders in general fit into the work of this committee and not just focus on the newborn screening aspect.

So this, again, is very preliminary. These are kind of initially what we discussed at our meeting, but I think we have more work to do in identifying what might be some of the priorities, where can we have the impact, and where can we not be repetitive in our work. And also, obviously, we would like to have input from the committee on new priorities and projects as well.

And let me just say before we -- does anybody from the committee wants to chime in on anything or clarify anything that I did not?

DR. BAILEY: No. Just to reinforce and, first of all, thank you for taking over as chair of the subcommittee. I am personally grateful, most grateful. But,

obviously, Catherine did a great job in the meeting yesterday in (inaudible) so the committee have been excellent.

The point that I've tried to make in previous meetings, and that you made very well here, is that the education and training space is very big, and the needs are unlimited. So the question -- and there are a lot of players in this already. We've got the -- you know, the professional organizations having (inaudible) standards or whatever, or, you know, whether it's pediatricians or family practice people, really don't want to try to change those necessarily -- developing training curricula is not really our responsibility.

Are there some, you know, recommendations we should be making? (Inaudible) looking for guidance from the larger committee and --

CHAIR BOCCHINI: All right. Thank you. (Inaudible) yesterday. Let's open this up to the committee and to the --

PARTICIPANT: Well, going back to your Priority A and your -- the discussion you had about where screening in childhood fits, and whether that is really a clinical practice guideline or a public health issue. It parallels somewhat with developmental screening, which certainly in terms of autism is in the public health domain.

And I think, you know, in our Title V program we are very much interested in taking steps in public health to support practitioners in implementing those clinical guidelines. So, yes, there are clinical guidelines around development screening, but a lot of -- a lot of practitioners aren't able or knowledgeable enough to do that yet.

So we see part of our role as supporting them to implement that, and I think maybe a parallel vision of how to do this -- do other types of training (inaudible).

DR. BAILEY: I would just respond to that and say that's very helpful, and I

think, you know, it's going to be an interesting and complicated paper to pull together. I think one of the things we are going to be focusing on is where we originally started out discussion is, would there be any value in later population screening? So move into (inaudible) screening; that's a whole different set of -- that's a whole different set of considerations. Have you got all this out in the paper? Is (inaudible)?

DR. THOMPSON: Cathy, you raised a really interesting point about where -- how much of our role is the diagnosis and early intervention for heritable disorders and how much of it is sort of the broader picture. And it strikes me that what you're bringing up I don't know necessarily fits in your committee, and I guess the question is, is there a need for an additional committee that looks at -- certainly, there are other clinicians at the table whose states are right now sort of reconfiguring how you manage complex chronic

illnesses.

And there are some real concerns that those children, based on decision-making, may very well be put into care systems that really actually don't favor them, for instance, being seen by the specialists who can take care of them best, because they're aligned with their primary care provider.

And so it strikes me that many of the children that we are identifying as children who have heritable disorders may find themselves very quickly no longer actually being in a position to actually receive the highest quality, evidence-based care.

I guess the question for me is, is that something that is under the interest or domain of this committee? And if it's not, is that something that the committee -- something that has to do with access issues, health care financing. I mean, there is -- so these broader issues, you know, if we are looking toward these children being providers, they

won't -- in all likelihood, they will be kids with chronic, complex problems.

And the question would be is, are we positioning their care to be structured in an evidence-based way to actually ensure that they actually have access to providers? And I have to tell you at this point, you know, we are -- we are quite concerned that there are a number of our patients that are already being referred elsewhere. And it's a choice of which Medicaid program does that (inaudible) to. So, and they get to keep their pediatrician, but that they have no access to actually specialty care.

And it's (inaudible) and really right now it's -- and I can only assume at this -- and so do we have an opportunity to be -- I don't know that it necessarily all falls to us. I'm just wondering, is there an opportunity for this committee to examine what, if any, role we have in this, since we -- it's fairly clear that this is -- the train is leaving the station. I really don't see us going backwards. I see

this as being --

CHAIR BOCCHINI: So, Carol?

DR. GREENE: I would like to passionately second what Alexis just said, and to say that -- and Coleen will certainly tell you that that has been a discussion among the clinical parts of the committee from the time that she was chair of the Follow-up Committee, and a much better description of what I was heading for in the discussion of the Long-Term Follow-up Committee, wanting to look at access issues. All sorts of things have come up before, but that -- that I have always felt, from the time Coleen was chair of the committee, previous committees, I mean, that is actually in the charge of the Long-Term Follow-up Committee.

And the reason that some of us are hoping that the Long-Term Follow-up Committee looks not just at the data that tells us, are we doing it and what has happened, but actually looks at the issues themselves and tries to help

the committee, which I am only a liaison, but that the -- one of the roles of the Long-Term Follow-up Subcommittee -- and I personally would not want to see a separate implementation committee, because, you know, you can implement the lab, you can implement everything.

And I think directly in the charge of the Long-Term Follow-up Committee is that people need to have treatment. They have to have high-quality treatment, not just access to treatment, they need to have treatment. That's what long-term follow-up is. And I think that's directly in the charge of the Long-Term Follow-up Committee.

PARTICIPANT: I agree.

CHAIR BOCCHINI: Okay. I have you, (inaudible), and then --

PARTICIPANT: Thanks. I just very briefly wanted to bring up the idea that in conjunction with expansion of newborn screening is the expansion of prenatal screening. And from my vantage as an

obstetrician, there is a whole bunch of effort and, you know, expanded carrier screening. So people are coming into newborn screening with a whole lot of stuff that has already gone on.

There is a couple of issues there. One is education for the parents and clinicians, how to get that information from the prenatal period into the newborn period and childhood period and siblings. What are the ramifications for siblings?

And there is also a (inaudible) for an information systems piece. Is there a way to do that?

But I would just want to throw it out there, something that could at least just be noted, because from a patient perspective and from the continuum of care, I think we have to try to remember that piece if we really (inaudible) that we, as clinicians, go on the prenatal -- the post-natal side, but of course families are moving through this. So we want to try to integrate that education perspective.

MS. WICKLUND: I don't have to be the one -- remember, I think last time I said that, and now you get to keep saying that.

Let me just -- one of the things that I just want to just make a note of is implementation science in general, and what implementation truly means. So, and I am not an expert in this area, but one thing I have learned over and over again when I am around implementation people is that it is a science of itself that is multi-faceted and includes a lot more than, you know, so many different aspects than -- and one of the reasons why it's such a complicated topic to tackle, one in which we have tried in the Institute of Medicine Roundtable, and have kind of gone there, kind of backed away a little bit, gone there, backed away a little bit, but I just really want to make sure that if we are going to tackle implementation, if we want to, it truly is more than -- even some of the stuff that we are mentioning right now, it's just, you know,

huge. It's very complex.

PARTICIPANT: And along those same lines, I do think it would require a very different expertise than I would argue is currently at the table. It would seem that the individuals that come into that conversation come from very different backgrounds.

I also meant to mention when we talked about this -- the relationship to education and training, that in addition to having patients and not being certain where their care is, there continues to be, you know, I think the notion of it not being clear why a genetic counselor, for instance, is someone who actually needs to be part of a health care team, whether it's -- it's looking at reimbursement or whatever, the notion that training is tied to actually being able to provide that kind of care and really being able to clearly define what does a genetic counselor do, whether pre-natal or post-natal, but to sort of make it clear that as we're trying to look to where does

training meet long-term follow-up or even, you know, last standards that somehow we can be clear on why that particular -- is not obvious to some people.

DR. HOMER: But I think it's actually fair to say that Dr. Daugherty and I probably are (inaudible) experts in implementation science. So there aren't a whole lot of us out there, so I'll take out my card.

So I think what I'm hearing from at least members of the Long-Term Follow-up Committee is some sense that perhaps we are not fully addressing some things that -- which I tried to put on my final slide. But I do take that under advisement and think basically we will bring that back to the committee.

I think -- I mean, a broader question is, to what extent are we an advocacy group, or to what extent are we seeking to identify data, gaps in data, and point that out. And that is I think really what I have been

wrestling with as chair, and so, again, that may or may not be a -- that may be an offline conversation. But that's, I think, partly what (inaudible) through here.

CHAIR BOCCHINI: I think that's really -- this is a good discussion. You know, we are policy committee. We're -- so I think that our goal is just as you said, Charlie. We need to try and use the expertise on this committee to develop the best policies for the health of children and families. And then the goal is to recognize absent that -- or provide opportunities to be able to study how that is happening. But the information I think is not a key part of this committee -- think we can -- on what is not happening.

And I think the framework that was developed by the Long-Term Follow-up Committee about picking a condition and sort of looking at ways that people could look at the effectiveness of the long-term follow-up and treatment is -- is appropriate for this

committee to do. But whether we should be able to go into actual -- make recommendations for how to implement, I think it's -- I mean, we can debate that further and discuss it further, but I -- and I think that's -- so, Don, and then Carol.

DR. BAILEY: Yes. That's a helpful -- helpful comment. I think the same applies to the Education and Training Committee is what is -- what is the role of the committee, and if it's around policy as opposed to -- keep in mind.

One thing that strikes me from this conversation is that, you know, we are three subcommittees operating somewhat independently. We do come together and talk in our -- in this big meeting, but clearly some of the work of the Education and Training Committee could follow on directly from the other two subcommittees.

And so there may be -- you might want to think about some kind of structure in future

meetings where we, you know, maybe have our separate committee meetings, and then -- actually, I don't know what the right structure would be there, but I think for us to hear from long-term follow-up, what are the top education and training priorities, same thing from education -- I mean, from the laboratory standards committee, is there anything we can do to help advance --

CHAIR BOCCHINI: We did have -- we have had phone conversations amongst the chairs of the three subcommittees to see where things were and to help us fertilize what was going on and --

DR. GREENE: The comment was made -- or the question was, and you have already answered, Joe, this is clearly not a place for advocacy. With that said, I think there are important ways that we can look at what is happening and explore -- and this is a term that has been around in the long -- the LTFU, Long-Term Follow-up and Treatment Subcommittee

for a number of years, and we have kind of been working up to looking at roles and responsibilities.

And there is this huge opportunity right now, because health care is enormously influx, and things are happening and some of them are very, very good, and some of them it's very clear are putting our special kids and families at huge risk. And you just heard Alexis describe some of that.

This is a time where if we look at those, not just the -- what is happening, but how it's happening, that we begin to look at the states, at the models, that doesn't get into advocacy, but it does get into understanding what is going on and seeing what the opportunities are.

And this is a time to be able to look at those roles and responsibilities, what states are doing, how people are making sure that people do have access, how people are losing access, and that is a place where I think

this committee can -- has an opportunity to make a huge contribution in a very timely fashion, and then hand that all over to the people who do advocacy.

I guess I'm thinking of Alexis' expertise that is missing. Thank you for letting me join your club.

PARTICPANT: I think some of what you were saying is something like Carol was just saying, that the health care delivery system (inaudible) changing dramatically -- kids who might have been able to go to a children's hospital may not be able to go there anymore, that kind of stuff.

And that kind of financing -- knowledge and where the policy letters are and whether this committee is able to say anything about -- that's the kind of thing I -- I know it a little. You know it -- you have to know -- access to quality.

But -- I was on -- I used to be on the interagency Autism Committee. That

committee actually wrote a nicely worded recommendation to the Secretary, but it was done with a lot of input from somebody from CMS who knew a lot about certain (inaudible) rules are. Those people are rare, you know. Everything about the Medicaid rules --

Anyway, does that -- am I getting at what you were --

MR. OSTRANDER: Robert Ostrander, New York City Academy of Family Physicians and NYMAC. I got involved in this world a long time ago and got into this through a circuitous pathway by participating in one of the first NICHQ medical home learning collaboratives, Charlie.

And I learned about what a patient- and family-centered medical home was then. And I look at what the NCQA calls a patient-centered medical home now. I kind of prefer payer-centered medical home myself, if that's not too politically incorrect.

And what I want to point out -- and

this really just is -- I'm just saying a different way what Carol already said -- is that the evolution of health care and this grand plan that we have as a nation to bring everybody under the umbrella has resulted in a system that ignores the exceptional patient in order to save the money and provide lots of health care to the folks with common lifestyle problems.

And so systems are put in place, and they are rigid systems, to either incentivize or require certain systems of care that don't work very well with the original patient- and family-centered medical home, which involved personalized care plans and not disease-based care plans.

And although I don't think we can advocate here -- I understand that, I have a hard time not doing that, but I understand that -- I do think that we can identify barriers. And if you identify barriers, we are not advocating until we say what the solution is.

And I think, you know, this is

honestly where we really are in a full circle on the Follow-up and Treatment Subcommittee, because the reason we set up this framework was to help identify barriers to the paper we wrote before about what the key elements were for long-term follow-up and treatment of folks with special -- with heritable diseases. And that involved a lot of this kind of softer, non-measurable stuff like, are they in a patient-centered medical home, and what does that mean for somebody.

So I really do think it's our purview, and I would love to -- at least for part of what we do in the Follow-up and Treatment Subcommittee, is to pull back a little bit. I mean, we did a lot of numeric stuff this time, but pull back a little bit and actually talk about what follow-up and treatment looks like.

Transitions is huge. We are doing stuff in NYMAC where -- and, again, we're ready at ground zero. It's very clear to me that primary care folks in -- and, frankly, on both

ends people don't understand what transition is all about. This came, again, out of that first NICHQ initiative all those years ago when I got kind of excited about transitions.

There is no education. People don't know what it means. They don't know what it means at the subspecialty end. They don't know what it means in primary care. And in NYMAC we're starting with just a simple survey of the training programs to see, is there any training in transition? Everybody who becomes a pediatric subspecialist has to be a pediatrician first. Everybody who comes in as an internal medicine subspecialist has to be an internist first.

And I think that those things really very much are in our purview. I mean, we can identify barriers, lack of education and transition. There are barriers in the way the health care system has evolved that have been wonderful for the nation as a whole, and they're getting more people taken care of and more

people insurance.

But the fact that there is lack of a system to identify exceptions and waivers, however way you want to phrase it, so that our -- so that these folks can stay with the care providers that can best serve them if you meet X, Y, and Z criteria. Those are the kind of systems processes that I think we can drive, so I would very much like to -- and I don't think we need another subcommittee for that. I would, you know, very much like to see it be part of this committee as a whole's work, and also our subcommittee.

CHAIR BOCCHINI: Additional questions?

PARTICIPANT: I have a comment that's a little bit off topic, but this -- I'm not sure whether the Education and Training Committee is the right group to tackle this, but it seems to me like there is an opportunity for education around later onset disease (inaudible). I just wanted to throw that out

there as -- as a topic that the committee may --

CHAIR BOCCHINI: Any additional comments? Again, now we have just -- next is to discuss future topics, and I think (inaudible). And any other topics? I know Steve -- but he did mention a potential topic for us, one that -- significantly I guess is the FDA has made a decision about kits for -- help me explain this, but I guess it has to -- but it has to do with home --

DR. KELM: Yes.

PARTICIPANT: -- for subsequent genetic testing and --

DR. KELM: We have notified Congress that we intend to release stress guidance, although our notification included a draft of that guidance. So that indicates a framework for regulating laboratory development tests. We would not force anybody to use (inaudible), but it would just be the fact that some of the -- you know, that we would assess what is on the market, because honestly

no one knows, and then deciding -- start with probably the most risky (inaudible) and asking them to come in (inaudible) FDA for clearance approval.

And there are exceptions in there for rare diseases and some others, although I know from talking to Mike Watson previously he wanted to know what that -- what that meant, what was the definition for that. And I agree that will be a good definition to add. But I'm sure that that probably be a discussion that we would interested in.

CHAIR BOCCHINI: Okay. All right. So that would be something that -- thank you for clarifying that for me.

All right. Additional potential topics? Don?

DR. BAILEY: So I know that at a previous meeting I believe Tiina had gave a brief overview of the four centers that were funded, to look at whole genome (inaudible) sequencing. Those centers have been up for

about a year now, and maybe by the -- either the next meeting or the next meeting, it might be interesting to have a report. There's a lot of debate and discussion about whether that is even a good idea.

I think -- being a part of one of them, I think the research piece of it is very important work to be done to help inform future -- future policy. So at some point I'd be glad to organize a session where we would give an update on kind of what questions are being addressed by what -- how the project has evolved over the first couple of years and where we're --

CHAIR BOCCHINI: Steve, and then Carol.

DR. McDONOUGH: Where I work in Bismarck we have had several children who presented with critical congenital heart disease that had a normal O2 sat screening in the hospital. And these children have a condition called coarctation of the aorta,

which is not one of the original seven or so that were designed as the O2 sat we were screening.

I think that providers may believe that the O2 sat screening are picking up the most critical heart diseases. And sensitivity on a -- publication, I was looking at a lot of studies last year, indicated sensitivity is about 75 percent.

So, as a pediatrician, I would like to know what the American Heart Association and American Academy of Pediatrics would recommend should we have children come back at three days of age or seven days of age, and should -- screenings on them, see if we can pick up some of these children with coarctation before they present in congestive heart failure or shock.

This would not be something we would be asking the health department to do. You know, it's the recommendation for primary care providers. Is there something we can do to pick up these kinds who are being --

DR. GREENE: Before I get to my

suggestion, I'm pretty sure, but I hope I'm not misspeaking, to follow up on what was -- Steve just said, I'm pretty sure that coarcat is not a cyanotic disorder, and that repeating that screen won't pick up coarcat. So that if it's going to be an important issue for education, I will be interested to know what people do for education.

But that -- that HD screening does not pick up anything that's not cyanotic, and I think even bringing them back to -- the screen won't pick it up.

Yes. So it's -- what I was wanting to bring forward might be related to what Alexis, what we were just talking about about the whole models and access and understanding and not advocacy. But we have had -- I think it was -- Debi Sarkar, you will know for sure when we had a presentation, and at that time it was sort of focused on the ACA and everything was very, very politically sensitive. It was just coming in.

And we had a presentation -- the committee had a presentation on the ACA, and I believe the subcommittee did as well. And it was pretty 30,000-foot, this is how it's going to work, and it was really focused on the healthy kids.

There was nothing in the presentation -- Alexis at that time I think did a presentation about some of the concerns, but the people who presented about the ACA and about the new systems and about -- and it's now more complicated really did a -- this is how wonderful it is going to be, that kids are going to all have primary care and coordinated care.

And I think it would be a good time to have some really thoughtful presentations about what is happening as it is being implemented to -- we keep saying, and we know from our own experience that the children with the rare, complex disorders, the children and families are being hurt by some of the changes. And not to point fingers, but just to understand

what's going on, I think it would be wonderful, at least at the subcommittee and I think probably at the committee level, to really dig in and have some presentations about how that is working, so we can begin to understand and begin to understand what we should be looking at, and not -- you know, not just at this -- how the ACA works and how you get you primary care, but what are truly the impacts that people like Alexis and I and Debbie Badawi are all dealing with when we try to provide the care that we know is needed.

CHAIR BOCCHINI: So I want to thank everybody. I think we've had a really excellent meeting. I want to thank Debi for organizing this and setting up the agenda, so that it has been very, very -- very well done, and I want to thank everybody for their contributions. Certainly, I think this is good evidence that a face-to-face meeting -- all for your participation. Look forward to seeing you again in February.

MS. SARKAR: The next meeting is February 12th and 13th.

(Whereupon, the above-entitled matter went off the record.)