

1 **SECRETARY'S ADVISORY COMMITTEE ON HERITABLE**

2 **DISORDERS IN NEWBORNS AND CHILDREN**

3 **Friday, January 28, 2011**

4 **Post mid-morning break**

5 CHAIRPERSON HOWELL: We're going to hear a long-
6 awaited presentation. We're going to hear a long-awaited
7 presentation about the efficacy of newborn screening from a
8 family perspective. We ran out of time at our last meeting
9 to hear this presentation, so Dr. Kaufman was very kind to
10 return today.

11 Dr. Kaufman is Director of the Research and
12 Statistics at the Genetics and Public Policy Center at Johns
13 Hopkins University, and that center is based here in
14 Washington. He is responsible for the design and analysis of
15 the center's public and professional opinion surveys on a
16 range of topics, including newborn screening, direct to
17 consumer testing, and return of individual results to persons
18 participating in genetic studies.

19 In 2009, HRSA funded several studies to examine
20 the efficacy of newborn screening from a family perspective,
21 and Dr. Kaufman will present the results of one of those

1 studies. Dr. Kaufman.

2 I also might point out that we have Kaf on Skype
3 over here, so that not only can we hear him, but we can see
4 him. It looks like his photograph has now gone to about one
5 inch square. It was quite bigger earlier on that screen.
6 But anyway, you are being watched and we're watching you,
7 Kaf.

8 EFFICACY OF NEWBORN SCREENING FROM A FAMILY PERSPECTIVE:

9 A NATIONAL SURVEY OF RECENT AND

10 PROSPECTIVE MOTHERS ABOUT NEWBORN Screening

11 DR. KAUFMAN: I'll keep that in mind, that I'm
12 being broadcast.

13 DR. LLOYD-PURYEAR: To Ghana.

14 DR. KAUFMAN: Oh, wow.

15 (Slide.)

16 Thanks so much for having me. I really, really
17 appreciate it. You guys had really important discussions
18 going on last time, so I'm happy to be able to make it back.

19 (Slide.)

20 As a lot of you probably know, a lot of the
21 surveys that are done to ask folks what they think about

1 newborn screening are often done in parents of affected
2 children or in parents who are at high risk. Those are --
3 that's important work, but HRSA thought it might be
4 interesting to find out what sort of the general public sort
5 of knows and thinks about newborn screening, and as part of
6 the project that we did with the Genetic Alliance and the
7 folks at Maryland we conducted such a survey.

8 So the goals of the project were to measure the
9 general public's knowledge and understanding of newborn
10 screening, to see whether people support the general ideas of
11 screening, to see what sort of information people are getting
12 and would like to get, and to sort of examine as you guys
13 think about expanding panels, what the public thinks about
14 prioritizing, what kinds of diseases they would like to have
15 screened. So we tested three factors: severity of disease,
16 age of onset, and the positive predictive value.

17 (Slide.)

18 I'm so sorry. I have a pretty annoying cough, so
19 I'm going to go with a cough drop here, which means no
20 hugging or kissing afterwards.

21 We conducted the survey on line. It took about 11

1 minutes on average for people to take the survey. We used a
2 national, random sample of two groups of women that we got to
3 through our survey partner, Knowledge Networks. I can talk
4 more about their panel at the end if you have questions.

5 So we wound up talking to about 2200 women between
6 the ages of 18 and 45. There were two groups: recent
7 mothers who had given birth in the past three years; and the
8 group we called prospective mothers, who said they were
9 planning to have a biological child in the next three years.
10 Of course, we know that that's an imperfect measure, that
11 plans don't always go according to plan.

12 But we thought it would be good to talk to recent
13 mothers in order to get perspectives on recent experiences
14 without having too much recall bias, and then also find out
15 what a group who hasn't had the experience recently knows.

16 We asked all the women 46 questions and then the
17 recent mothers got another 10 questions about their last
18 experience. Then we randomized people to view one of four
19 different testing scenarios, and I'll talk about that in a
20 minute.

21 (Slide.)

1 These are just the demographics of the two groups
2 of women. Fairly similar to one another, although
3 prospective mothers are a good deal younger. We don't need
4 to spend a lot of time there.

5 (Slide.)

6 So, what do people know about newborn screening?

7 (Slide.)

8 At the beginning of the survey, we asked: "Before
9 today, had you heard of newborn screening?" Only two-thirds
10 of the recent mothers and 38 percent of prospective mothers
11 had even heard of newborn screening before.

12 (Slide.)

13 This awareness was quite different according to a
14 couple of key demographic factors. As you can see, in both
15 recent mothers and prospective mothers awareness increases
16 significantly with education. That's a 30-point difference
17 between people who haven't finished high school and people
18 with a bachelor's degree. Hispanic women were significantly
19 less likely to have heard of newborn screening than white and
20 black non-Hispanics. It didn't seem to really vary that much
21 with respect to age or income.

1 (Slide.)

2 Asking the recent mothers about their awareness of
3 their last experience, 39 percent said that they had gotten
4 too little information on newborn screening, two-thirds said
5 they knew their baby had been screened at the time, 44
6 percent remembered getting the results of newborn screening,
7 and about a quarter said that they had a good understanding
8 of the process.

9 (Slide.)

10 We asked recent mothers to sort of just describe
11 and assess the amount of information that they got during
12 their last pregnancy. For in ten women said that they
13 weren't given enough information or they weren't given any
14 information. About 37 percent said they got about the right
15 amount.

16 (Slide.)

17 We asked the women, recent mothers, how well they
18 felt they understood newborn screening based on the
19 information that they've gotten to date. 80 percent said
20 they had a good or basic understanding. 20 percent, 21, said
21 they had very little or virtually no understanding of the

1 process.

2 (Slide.)

3 Then we asked both groups of women, if you were
4 having the child, another child, when would you first want to
5 get information about newborn screening? You can see if you
6 add those first four groups up that about 85 percent of
7 recent mothers and 90 percent of prospective mothers want to
8 get information on newborn screening before they get to the
9 hospital.

10 Compare that -- so those are the responses from
11 the recent mothers about when they would like to get it, on
12 the left in purple. On the right is when they actually got
13 newborn screening in their next pregnancy. So 55 percent of
14 women either got information in the hospital or only received
15 information if there was a problem. I think that's maybe one
16 of the more significant findings. I'm sure it's not a
17 surprise to you, but it does reiterate one of the issues of
18 getting information out; it's not getting there soon enough.

19 (Slide.)

20 We asked both groups of women what kind of
21 information would be the most important to them. Recent and

1 prospective mothers were pretty similar on this, so we just
2 combined them. Women say they want a lot of the practical
3 information -- what happens if the screen's abnormal, what
4 should I expect, what conditions are being screened for, how
5 will I get my baby's results, and what are the risks
6 associated with having the sample taken.

7 (Slide.)

8 Right after we gave women the definition of
9 newborn screening, we asked them what they thought of the
10 idea.

11 (Slide.)

12 There's very, very broad support for screening for
13 conditions where early diagnosis could improve the baby's
14 health. Almost all women, 98 percent in both groups, agreed
15 with that. Significantly fewer, but a large majority,
16 support screening for conditions where early diagnosis might
17 not improve the baby's health, and about 5 percent of women
18 said they felt newborn screening should never be done.

19 (Slide.)

20 We asked about some of the goals of newborn
21 screening. Over 95 percent in both groups said that it's

1 important so the parents can prepare to care for the child
2 with a condition; similarly high to improve the health of the
3 babies; and 88 percent of prospective mothers and 85 percent,
4 so they can learn if they're at high risk to have another
5 child with a condition.

6 (Slide.)

7 The greatest concern about newborn screening was
8 the worry that it might not provide accurate information,
9 nearly half of women; lesser concern about it causing too
10 much anxiety and taking money away from other health care
11 needs.

12 (Slide.)

13 Sort of as a lark, but also just to see, we asked
14 women if they'd be interested, if such screening were
15 available, tests were available, would they want to screen
16 their babies for late onset, adult late onset diseases, and
17 for a bunch of traits. As you can see, majorities were
18 interested in screening for diseases like diabetes, heart
19 disease, colon cancer, and Alzheimers. We through type 1
20 diabetes in there as a control, explaining that that was
21 childhood diabetes. Similarly high interest in adult

1 diseases, then a fair smattering of people interested in
2 testing for traits as well.

3 (Slide.)

4 What about those three factors and how they
5 influence people's opinions about whether a disease should be
6 screened for or not?

7 (Slide.)

8 As I said earlier, we randomized the women. Each
9 woman saw one of four different scenarios, where we described
10 a fictional unnamed incurable rare genetic disease, and we
11 told half of the women that the symptoms appear late, between
12 12 and 15 years of age, and half of them that they appear
13 between 3 and 5 years. Then we told -- half the women saw a
14 scenario where we told them there's no cure and the disease
15 causes death; and half where it was a chronic disease that
16 gets worse but doesn't cause death, manageable symptoms.

17 (Slide.)

18 Each woman -- there were two versions of each of
19 those two things, so two by two. There's four scenarios.
20 For the scenario where women saw, we explained 90 percent
21 positive predictive value in some simple words and asked them

1 if they would support having all babies screened for that
2 disease. Then we said, now assume the positive predictive
3 value is 60 percent and asked it again.

4 (Slide.)

5 So which of the factors was most important? Well,
6 support for screening for a disease varied most when the
7 positive predictive value changes. So this is what recent
8 mothers said, and the dark blue is for a 90 percent positive
9 predictive value and the light blue is 60 percent. You can
10 see that no matter what scenario they looked at there's about
11 a 10 percent, 10 to 13 percent difference in support for the
12 screening if the positive predictive value is higher.

13 (Slide.)

14 What about the other factors? The other factors -
15 - age of onset and severity of the disease really don't make
16 that much difference. In recent mothers, on the top, you can
17 see if you start with late onset, chronic disease, support is
18 very similar for late onset fatal and for early onset chronic
19 diseases. In recent mothers, the situation where there's
20 early onset fatal diseases, screening for early onset fatal
21 diseases, support for screening is higher.

1 If you look at the prospective mothers, it's lower
2 for that. That's the only one that there's a difference for
3 as well, but prospective mothers are less likely to want to
4 screen for early onset fatal diseases. It's more of an
5 interesting sort of factoid than maybe that important, but
6 it's sort of interesting.

7 (Slide.)

8 So just in conclusion, the public needs and wants
9 information on newborn screening. Awareness is very low.
10 There's too little education and it's occurring too late, at
11 least according to the women we talked to.

12 I read in one paper that recognizes that education
13 is a problem and a lot of prenatal providers say that they
14 don't give information because patients don't ask about it.
15 But what we see is that women don't know about it to ask
16 about it. So if there's unfamiliarity patients won't ask,
17 and so doctors need, I think, prenatal providers need to be
18 more proactive.

19 Women want concise, practical information, and
20 they want it offered at multiple junctures during their
21 pregnancy, before they get to the hospital. And Hispanic

1 women, those under the age of 25, and those who haven't
2 completed college are all probably in greater need of
3 information efforts.

4 The goals of newborn screening are viewed very,
5 very positively, even by women who have gone through it
6 recently. So I think support for it in general is high.

7 Accurate information is highly valued. Again, we
8 saw the biggest worry was about inaccuracy, and support
9 increases with positive predictive value.

10 Majorities were interested in testing for adult
11 onset diseases and there is low but measurable interest in
12 testing for traits.

13 (Slide.)

14 Some of the limitations of our study. What people
15 say in a survey doesn't always correspond with what they will
16 do or what they really, really want. We did only conduct the
17 survey in English and so what we see in the Hispanic
18 population, lower awareness, is maybe even more pronounced
19 than what we actually saw, since we didn't talk to
20 predominantly Spanish speaking folks. A lot of the
21 variables, a lot of the things that we observed, do vary by

1 income and education, and so we want to go -- we should go
2 back and do more stratified analyses, although we did do a
3 lot of adjustment when we did regressions and what-not.

4 (Slide.)

5 I just wanted to thank our partners in this: the
6 folks at Genetic Alliance, Maryland, and Penny Kyler at
7 HRSA.a

8 Thanks a lot again for your time, and I hope it
9 was worth the wait.

10 CHAIRPERSON HOWELL: Thank you, David. Where were
11 these ladies located?

12 DR. KAUFMAN: This was a nationally representative
13 sample, so Knowledge Networks has a panel of about 50,000
14 people. They sent the survey out to 9,000 women across the
15 United States within the age group. They had to be eligible,
16 either as a prospective or recent mother, but it was a
17 national survey.

18 CHAIRPERSON HOWELL: Could you screen that, could
19 you pull out the Maryland mothers, for example?

20 DR. KAUFMAN: We don't have a ton of them, but
21 we'll do it.

1 CHAIRPERSON HOWELL: The reason I ask that
2 question, Maryland is one of the few states in the country
3 that requires informed consent, and it would be interesting
4 to see whether or not the mothers who gave informed consent
5 were aware they had newborn screening. I'd be interested in
6 that phenomenon.

7 DR. KAUFMAN: I'll take a look. We did ask a
8 question about, a couple questions about consent. About half
9 of the women felt that consent shouldn't be required, but
10 that parents should be able to refuse it, and about 43
11 percent of women said that they would like to be able to
12 choose what diseases their babies get screened for.

13 CHAIRPERSON HOWELL: Tracy.

14 DR. BLOUT: Dr. Kaufman, I worked in the Maryland
15 Health Department. We actually don't require informed
16 consent any more. They passed legislation in 2008 and we
17 just are requiring informed dissent. However, many of the
18 hospitals didn't know that and informed consents were still
19 included in educational materials, so a lot of hospitals were
20 still getting informed consent until this year. But
21 technically we don't any more.

1 CHAIRPERSON HOWELL: Thank you very much.

2 DR. KAUFMAN: Fortunately, our data's old enough
3 that --

4 (Laughter.)

5 CHAIRPERSON HOWELL: And most of us thought that
6 Maryland still required informed consent. But thank you for
7 clarifying that.

8 Tracy had a comment.

9 DR. TROTTER: Yes, I have a question. What was
10 the rationale for not having a scenario of infant onset with
11 the ability to have appropriate intervention with either diet
12 or therapy, which actually represents most of what we screen
13 for?

14 DR. KAUFMAN: The way we chose those scenarios, we
15 met with everyone on the project board and agreed that the
16 goal was to differentiate in areas where folks -- where the
17 folks thought there would be the biggest differences in
18 opinion, where the differences might actually really matter.

19 The fact that you -- Carol, can you speak to that?

20 DR. GREENE: I was one of the people who had the
21 good fortune to participate.

1 DR. LLOYD-PURYEAR: You should introduce yourself.

2 DR. GREENE: Hi. I'm Carol Greene at University
3 of Maryland, and University of Maryland was a very small part
4 of the project, but I had the very good fortune to
5 participate as he was doing this incredible job developing
6 the survey instrument. I think we shouldn't forget that the
7 fundamental part of the survey, 98 percent of women said that
8 they're in favor of newborn screening, and that was explained
9 as conditions that we treat.

10 So this was to look at other things beyond that.
11 So the whole of the survey to start with was based on the
12 basic definition.

13 CHAIRPERSON HOWELL: Gerry.

14 DR. VOCKLEY: Do we have or do you have any way of
15 knowing if any of those participants actually had literature
16 given to them sooner than they report? In other words, were
17 they handed something in a prenatal visit that just didn't
18 register, and then it wasn't until later when someone asked
19 them if they'd reviewed it or they were faced with a question
20 in the nursery that it actually came to their consciousness?

21 DR. KAUFMAN: No, we don't know that. I think

1 that what you're getting at is probably -- the fact that so
2 many women don't remember. There's probably quite a bit of
3 information handed out that doesn't register.

4 Now, almost everyone says, where do they want to
5 get information, first they want to get it from a nurse or a
6 doctor. Second is a pamphlet. Interestingly, I don't know
7 where you guys are in the process of education, but only 20
8 percent want to get it through the Internet. So is not where
9 women are looking for this kind of info.

10 CHAIRPERSON HOWELL: We have a major effort
11 emerging in education.

12 Thank you. Chris has a comment.

13 DR. KUS: Is there any way that you had to know
14 that any of the mothers had false positive tests?

15 DR. KAUFMAN: We did ask. It's a fairly small
16 number, I think less than 20. But we did go down that road
17 with recent mothers, what did you get, do you remember the
18 results, what were they, did it turn out your child to have a
19 condition. I think it was maybe a little more than that
20 actually, 30, 40. It's been a while since I've looked
21 directly at that.

1 DR. KUS: Were they positive about screening?

2 DR. KAUFMAN: Yes.

3 DR. KUS: The ones that had false positives?

4 DR. KAUFMAN: Yes, no difference.

5 CHAIRPERSON HOWELL: We will hear some very brief
6 comments from the audience, Ann, because we are going to stay
7 on time, for reasons I discussed earlier. We're getting a
8 little behind.

9 DR. COMEAU: Thank you. I have a different kind
10 of a methods question for this study and for other studies,
11 because this is not the first survey that I've seen that
12 utilizes Knowledge Networks. In developing the randomized
13 group, do you know whether or not any of these women
14 participated in other newborn screening questions, or was
15 this their first participation? I think that's very
16 important as we go forward.

17 DR. KAUFMAN: It is important, and we don't know.
18 It is important. We use them a lot and do serial surveys,
19 surveys of similar populations. In the VA we've done a
20 couple of surveys. It is important to try and at least
21 acknowledge that. I honestly can't speak to what this group

1 has seen.

2 Do you know when that other survey was done?

3 DR. COMEAU: I don't know.

4 DR. KAUFMAN: Thanks.

5 CHAIRPERSON HOWELL: Thank you very much. We're
6 going to zip along here, I think. People in the audience, we
7 have Kaf on Skype over here from Ghana, and the barking you
8 hear in the background I think is fresh from Ghana. So we'll
9 move ahead.

10 DR. FREMPONG: Thank you very much, Rodney.

11 CHAIRPERSON HOWELL: Oh, good, Kaf. It sounds
12 like you have a friendly dog with you there today.

13 DR. FREMPONG: No, there's no barking here.

14 CHAIRPERSON HOWELL: Oh, goodness. That does
15 complicate things.

16 (Laughter.)

17 Not only me, but others have heard the barking.
18 If it were only me, then that would be a bigger problem. But
19 anyway, maybe one of our other colleagues on this side of the
20 Atlantic has a dog. I hope all is well in Ghana today, Kaf.

21 We're going to zip along and hear about

1 overarching questions for newborn screening long-term follow-
2 up from Cynthia Hinton. Cynthia is from the Subcommittee on
3 Treatment and Follow-up, and she's going to be discussing the
4 committee's project to define the major overarching questions
5 to be answered so that newborn screening is meeting its goal
6 of achieving the best quality outcome for affected children.

7 Cynthia is a health scientist in the pediatric
8 genetics team at the CDC's National Center on Birth Defects
9 and Developmental Disabilities, whose acting leader is our
10 fine Coleen.

11 **OVERARCHING QUESTIONS FOR NEWBORN SCREENING**

12 **LONG-TERM FOLLOW-UP**

13 DR. HINTON: Thank you very much, Dr. Howell.

14 (Slide.)

15 This is a paper that we've been working on in the
16 subcommittee and have brought it before the committee as a
17 proposed statement from the committee.

18 (Slide.)

19 The purpose of this paper was to present broad
20 questions and important issues for consideration when
21 assessing if long-term follow-up is meeting its goal.

1 (Slide.)

2 A little bit of the context behind this paper.

3 About two years ago, the subcommittee met and they were
4 looking at issues more focused on variables. The NBSTRN had
5 started. CDC had a project. There were a number of HRSA
6 projects. At first the committee had thought maybe we ought
7 to make some recommendations about what variables in data
8 people should be collecting.

9 At the course of the subcommittee meeting, I
10 believe Alan Hinman had suggested perhaps the role of the
11 subcommittee is actually to not tell people what variables
12 should you be collecting, but to actually step back a level
13 and think, what are the questions that we're trying to
14 answer. There may be some different variables underneath
15 those, but that you're aiming for a very particular goal.

16 Also understand that with these national efforts
17 and regional efforts that are going on, there's been a lot of
18 collaboration. So we are in fact really working on
19 standardizing the variables that we're collecting and being
20 very aware of, on the IT level, what kinds of things are
21 being collected as well.

1 (Slide.)

2 So this paper built off of the previous work of
3 the Follow-up and Treatment Subcommittee which resulted in
4 the Kemper et al. that was published in Genetics in Medicine.
5 This was stating what the four goals of long-term follow-up
6 are, which is care coordination, evidence-based practice,
7 continuous quality improvement, and new knowledge discovery.
8 This came out as a statement of the Secretary's Advisory
9 Committee.

10 We were building off of what those goals were, and
11 we were looking at materials that were presented from the
12 regional collaboratives, ACMG, and NBSTRN and CDC projects.

13 We developed a one-day workshop with participants
14 that, if you have the paper, are listed in the back of the
15 paper, to talk about what would be the questions underneath
16 this goal that you would want to know. Then we have shared
17 this with other groups, such as the NBSTRN, regional
18 collaboratives, Long-term Follow-up Workgroup, APHL
19 roundtable, etcetera.

20 (Slide.)

21 This is the matrix. We have three audiences: the

1 perspective of families, the perspective of the medical home,
2 primary care provider, specialists and investigators, and the
3 perspective of the nation-state. Then at each of these
4 levels, we ask these different groups to think about care
5 coordination through a medical home, evidence-based
6 treatment, continuous quality improvement, and new knowledge
7 discovery.

8 (Slide.)

9 Of course, after the work group there was
10 brainstorming, and then a lot of reiterations about the
11 questions, until we finally came up with our final list of
12 questions.

13 This is care coordination through a medical home.
14 The overarching question from family would be: Is my child
15 receiving coordinated care through a medical home? Likewise,
16 for the providers and state and nation, are children,
17 adolescents, receiving care coordination through a medical
18 home?

19 Underneath that would be questions, still very
20 broad questions, but more specific questions that you would
21 collect information to answer, such as: Does the child have

1 a family-centered medical home? Is the family and knowledge
2 knowledgeable about the disease? What do families need to
3 facilitate following through with the treatment plans? These
4 are just some from the family perspective.

5 From the provider or investigator perspective
6 would be: What percentage of children identified through the
7 newborn screening program have an individual health care
8 coordination plan that is updated at regular intervals? And
9 then from a state or national perspective, you would want to
10 know what percentage of those children are in a family-
11 centered medical home? Do they have a care coordination plan
12 that is regularly reviewed? Are services available? Do they
13 have financial access to services? Have they transitioned to
14 adult care? Also, how many children are lost to follow-up?

15 (Slide.)

16 For evidence-based treatment, do they have a good
17 understanding of treatment regimen, how are children doing,
18 questions like "Are children identified through newborn
19 screening and enrolled in care doing better than those that
20 were identified clinically?" I think this has come up
21 specifically with expanded newborn screening and people are

1 just interested in knowing. We have a population of children
2 that were identified clinically before expanded newborn
3 screening and those that we're picking up through newborn
4 screening, and that's just to answer the question, are we
5 really making -- are we really making a difference in this
6 early intervention. Because these conditions are so rare,
7 this is just starting to build that evidence-based review for
8 the identification. At any rate, those are: Are children
9 doing well?

10 (Slide.)

11 Continuous quality improvement: Is my child
12 getting the best care and treatment? How can I improve my
13 child's outcome? So is up to date information on treatment
14 made available to families? What percentage of families feel
15 they have ongoing access to age-appropriate education? Do
16 families have the opportunity to be in communication with a
17 medical team for effective management of their child's care?

18 From the provider perspective: Is there an annual
19 review of best practices and care plan for each child across
20 all levels of the care continuum?

21 I do know that people were --

1 (Electronic popping sound.)

2 There's that dog again, the dog that's not in
3 Ghana.

4 We're still in a process of developing best
5 practices and care plan for each child. Again, these
6 conditions are very new and I think part of the ongoing data
7 collection process will actually help improve the development
8 of best practices and care plan. Then for states and the
9 national level, how do we assure ongoing quality and ensure
10 quality improvement?

11 Is there a coordinated ongoing process for
12 collecting and synthesizing information about effective
13 treatments? Is there a coordinated mechanism for connecting
14 affected individuals with the most effective treatments or
15 clinical research trials if the appropriate management is
16 uncertain? Is there ongoing evaluation of the effectiveness
17 of various treatment and protocols and regimes and are there
18 policies in place at the state and national level that
19 facilitate collection and exchange of information among all
20 components of the newborn screening system?

21 (Slide.)

1 New knowledge discovery really relates to clinical
2 trials, clinical research. So: Is my child able to enroll
3 in clinical research related to his or her disorder? What
4 percentage of families are engaged in the development of
5 disease-specific registries, standardization of best
6 practices and research studies?

7 Do children in my care -- this is from provider
8 perspective or investigator perspective: Do children in my
9 care have the opportunity to enroll in clinical research? So
10 a provider could look at what percentage of children are
11 enrolled in clinical research. Does the enrollment in
12 research influence outcomes? Is knowledge gained from
13 longitudinal studies informing clinical care and treatment
14 development for children with these conditions? This is
15 really getting at the efforts of NBSTRN, where you would want
16 to have that translation from practice to care.

17 What clinical and observational long-term follow-
18 up research efforts are being performed at the state and
19 national levels? So are high quality newborn screening
20 surveillance and tracking systems in place at the state and
21 national level? Do states use national data and standards to

1 collect data and link systems? Are safeguards in place to
2 protect the privacy of children and families enrolled in
3 clinical research? And are the results of basic clinical and
4 translational research incorporated into best practices for
5 the care of children? (Slide.)

6 Next steps would be to guide current and future
7 data projects to develop systems that incorporate measures to
8 address these issues. Even as we've been in discussion with
9 people, such as APHL roundtables or with regional
10 collaboratives and states, people who have been interested in
11 the terms as we've been developing this matrix, to take these
12 questions back to their states or back to their regions and
13 just have them in front of them as they are thinking about
14 the types of variables that they would want to collect to
15 assure that they're doing the best for their state's
16 population.

17 Develop specific and measurable indicators for
18 long-term follow-up care after newborn screening, for example
19 through NCQA. There have been some initial discussions with
20 NCQA using the matrix and questions developed that were
21 really interesting, and I think that taking some of these

1 specific questions you then actually have a framework to sit
2 and start thinking about what's our numerator, what's our
3 denominator, and really putting some teeth into these
4 overarching questions.

5 Then assure adequate resources to accomplish the
6 goals of long-term follow-up care after newborn screening, as
7 well as ensure continuing resources for long-term follow-up
8 care in the future. This really comes back to discussions
9 that we've had around the table, either in the subcommittee
10 meeting when we were talking about point of service or other
11 considerations that we make, is that as you talk about data
12 collection, you talk about ensuring quality, assuring access,
13 and I think the question is what resources are going to be
14 available. But this provides a framework to at least say
15 these are the goals, these are the questions we need to
16 answer, and what do we have in place to answer those
17 questions.

18 At any rate, that's just a presentation of the
19 paper that I believe is in your packet, and I'd like to just
20 extend appreciation to everyone on the subcommittee that
21 helped work on it. Thank you.

1 CHAIRPERSON HOWELL: Thank you very much.

2 Are there comments or questions about this
3 presentation? Some of the authors of this paper are around
4 the table.

5 DR. KAY JOHNSON: Hi. This is Kay Johnson. Am I
6 on audio now?

7 DR. HINTON: Yes.

8 CHAIRPERSON HOWELL: Yes.

9 DR. KAY JOHNSON: I just joined you a short time
10 ago. As many of you know, this is an issue that I've long
11 been concerned about, and I just wanted to say that I think
12 the work has been tremendous over the past few years and
13 making real progress. I'm not going to dwell on it a lot in
14 my presentation today, but I do think that the opportunities
15 that are created by the expansion of children's health
16 insurance are a tremendous support to the thinking about how
17 we continue to invent this both at those programs, Medicaid
18 and CHIP, as well as in children with special health care
19 needs programs, so that we build a system that supports all
20 of our families and children that's right, with these
21 children well integrated into the systems; it's critical it's

1 moving in that direction.

2 DR. LLOYD-PURYEAR: I have a -- this is to Coleen
3 -- Executive Secretary question. Where do you see the
4 subcommittee going with this and how far are you -- what is
5 the role? Have you guys thought about what the role is of
6 this committee for going further? And is there more of a
7 role of a federal agency funding some of this stuff? Just
8 bureaucratic questions for the committee, for you, for the
9 committee to consider.

10 DR. BOYLE: I think the subcommittee we haven't
11 had that discussion yet, but I think those are great
12 questions, Michele. We've started down the path of NCQA
13 helping us with some guidance around measurement and next
14 steps. I think that might be the next step for us, thinking
15 about, again, trying to use the power of the committee to try
16 to help advance matters.

17 DR. KUS: I thought we were done. I thought we
18 did the paper.

19 (Laughter.)

20 But I think what this brings up again, and I think
21 that Kay's comment is correct, is, as we've said all along, a

1 system of long-term follow-up doesn't have funding. There is
2 funding to develop some things, but there's not ongoing
3 funding to collect this information. This is really to
4 stimulate the discussion about what could be collected. Then
5 we really have to say, how do we do this.

6 CHAIRPERSON HOWELL: Thank you very much. I think
7 that they're very good presentations.

8 So we'll move along and we'll go to our next
9 thing, which is health care reform and the implications for
10 genetics. We will hear from Kay Johnson and Brent Ewig.
11 Kay, who is joining us by telephone, has become very widely
12 recognized for her work in maternal and child health and
13 Medicaid policy development.

14 Do you have a comment?

15 DR. BOYLE: I have a question.

16 CHAIRPERSON HOWELL: A question, excuse me.

17 DR. BOYLE: I would like to know what our next
18 steps are with this. Can we submit it?

19 DR. LLOYD-PURYEAR: That's right. That's the
20 other bureaucratic question, because I received -- the
21 committee has to formally endorse this paper.

1 DR. BOYLE: As a product of the committee.

2 DR. LLOYD-PURYEAR: As a product of the committee.
3 It was sent out for comment. There were no comments
4 received, no dissension.

5 DR. BOYLE: There were a lot of comments on it.

6 DR. LLOYD-PURYEAR: Positive, yes. Nothing
7 negative. No changes, no changes, or nothing substantive.
8 So I think somebody needs to offer a vote to send it forth.

9 CHAIRPERSON HOWELL: Now it's been commented on,
10 again positively, as Coleen pointed out. So would you like
11 to recommend that it be sent forth for publication?

12 DR. BOYLE: I recommend.

13 CHAIRPERSON HOWELL: Is there a second?

14 DR. TROTTER: (Raises hand.)

15 CHAIRPERSON HOWELL: Those favoring the
16 recommendation?

17 (Show of hands.)

18 CHAIRPERSON HOWELL: Are there any nays on the
19 telephone?

20 (No response.)

21 There are no nays on the telephone, so it's

1 unanimous. So it will go forth. Thank you very much.

2 I was just telling you a bit about Kay Johnson and
3 her work in maternal and child health; and Brent Ewig, who
4 has arrived in the flesh, is Director of Public Policy and
5 Government Affairs for the Association of Maternal and Child
6 Health Programs, AMCHP, and in this role he works to advance
7 maternal and child health programs by assisting in the
8 development and implementation of public policy and
9 government affairs strategies, etcetera.

10 So this morning we're going to, at least on my
11 schedule, we are going to first hear from Kay, who's going to
12 discuss financing and status of reimbursement for genetic
13 services. Kay, you're there?

14 HEALTH CARE REFORM AND IMPLICATIONS FOR GENETICS:

15 FINANCING AND STATUS OF REIMBURSEMENT

16 FOR GENETIC SERVICES

17 DR. KAY JOHNSON: I am.

18 CHAIRPERSON HOWELL: Good. And your slides have
19 appeared.

20 DR. KAY JOHNSON: I apologize for having to join
21 you by telephone. I did have a prior commitment to be in San

1 Diego working today. But I am happy to join you by
2 telephone, and I know that having Brent there will give you
3 another anchor. Brent and I do presentations together from
4 time to time and I'm glad that he's there to complement and
5 add to the things that I'm going to say.

6 CHAIRPERSON HOWELL: We actually know that you
7 went out there to escape the snow, but we'll avoid that
8 comment.

9 DR. KAY JOHNSON: I was going to say, it's going
10 to be a lovely 75 degrees here today. Sorry.

11 (Slide.)

12 So I started thinking about this question really
13 at the request of Michele Puryear, who knew that I was
14 thinking a lot about children and health reform and women and
15 health reform and families and health reform, and began to
16 look more closely at what does this mean. Obviously, there
17 are a lot of unanswered questions about health reform and
18 some of those we'll come to later in terms of the politics.

19 But we do know some things from what's in the law,
20 and I think there are some opportunities to think about this
21 creatively and optimistically, as well as a few cautions.

1 Are you folks advancing slides on that end?

2 DR. LLOYD-PURYEAR: You have to tell us.

3 DR. KAY JOHNSON: Okay. I just wondered if you
4 were showing them. I am moving on to the slide that's
5 basically the first text slide, that says "The Affordable
6 Care Act builds on today's system and policies."

7 (Slide.)

8 DR. LLOYD-PURYEAR: So if you say "Advance Slide"
9 or "Next Slide."

10 DR. KAY JOHNSON: That's where I am, and then I'll
11 say "Next," okay.

12 So really thinking about the framework that this
13 falls into, one of the things that I think is so important,
14 as I said before, when we think about these children is that
15 they are part of the larger child health system. The more
16 that they are thought of as an important group within that
17 context, the more I think we assure their adequate health
18 coverage, their appropriate delivery systems, and so forth.

19 So what takes place -- the Affordable Care Act is
20 really building on what in GINA, on all of the advances in
21 Medicaid and CHIP coverage, and that ranges pretty widely

1 from Vermont, where we've essentially -- Vermont and
2 Illinois, that have made commitments to cover all children,
3 in essence, and in Texas, where they still have potentially
4 one out of every five children uninsured, and then a lot of
5 states that are moving rapidly with expansions under the
6 latest reauthorization of CHIP.

7 We're also building on a medical necessity
8 standard in Medicaid's EPSDT program, which is a child health
9 benefit for Medicaid and has a broad standard of coverage for
10 these children in terms of the array of services of specialty
11 care that they might need. We're building on the federal law
12 that will prohibit Medicaid and CHIP from denying children
13 coverage based on their health status, and that goes all the
14 way back to 1996, obviously building on all of the network of
15 things that have gone on in newborn screening and genetics
16 programs.

17 Next slide, please.

18 (Slide.)

19 I also think that we're reminding ourselves always
20 that there are many groups of children who have conditions
21 that we call by a lot of different names and that we slice

1 and dice in different ways. Sometimes these divisions are
2 driven by advocacy, sometimes they're driven by federal
3 funding, sometimes by professional units of service, so those
4 who care for one group of children are a different group of
5 professionals who care for a different group of children.

6 But I put this slide up here to remind us that we
7 have all these variations in how we talk about children who
8 have different kinds of special health care needs, the
9 children with heritable disorders among them and maybe even
10 really the majority of them.

11 So when we think about this larger context, it's
12 going to be always very important to frame it in this larger
13 group of children, I believe.

14 Next slide, please.

15 (Slide.)

16 I think in our individual arenas and for the
17 Advisory Committee as a whole, taking the word "advocate"
18 quite loosely and thinking about promoting, but using the
19 leverage that we have, we have much greater leverage if we're
20 not slicing and dicing up the children, so that those with a
21 physical versus a developmental disability, or those who have

1 an illness or a chronic condition versus some physical
2 disability, and those who have their conditions detected
3 through newborn screening versus other detection.

4 Again, thinking about them in a more unified way
5 and taking positions on benefits for that broad group of
6 children gives us an opportunity.

7 Next, please.

8 (Slide.)

9 I' going to frame my remarks in a health care
10 reform taxonomy that was developed by Sara Rosenbaum and her
11 colleagues at George Washington University. This was
12 initially developed for people to analyze the various health
13 reform proposals. It turns out to be equally useful for
14 thinking about the context of the legislation.

15 Next slide, please.

16 (Slide.)

17 So you see here the taxonomy itself, and I'm not
18 going to go over all of these items, but I think that you can
19 get the general sense that it's just a way to frame different
20 elements of the law. So whether you're talking about
21 accessing costs of coverage and consumer protections is

1 number four, over to public health investments, is number
2 seven, and I am going to go through a number of these.

3 Next, please.

4 (Slide.)

5 What does the Affordable Care Act mean in terms of
6 genetics if we're thinking about accessibility and
7 affordability? First of all, just the idea that we're going
8 to make coverage more affordable for more individuals, and
9 that will include some more children, although a smaller
10 number because so many of them already have gotten affordable
11 coverage under Medicaid and CHIP. But the idea that we have
12 young adults staying on their parents' health care plans
13 until age 26, and clearly the transitions for children with
14 heritable disorders into adults is a big issue and this will
15 affect many of them potentially.

16 The limits on lifetime maximums as well as the
17 prohibitions on health plans from dropping those who get sick
18 are obviously very important to these children, because even
19 though there were prohibitions put in in 1996 related to
20 children and heritable disorders or things detected in
21 newborns, they were not well enforced or strong enough.

1 Then that the law assures access to affordable
2 coverage for those now who are uninsured with preexisting
3 conditions through the short-term temporary high-risk pools
4 over the next few years until the law is fully implemented.

5 Next, please.

6 (Slide.)

7 I'm not going to read this whole item by Judy
8 Palfrey, but just thinking about the importance of not
9 denying coverage for a pre-existing condition for a child,
10 and probably everyone in this room having knowledge of some
11 experience where a heritable disorder or a disorder detected
12 in a newborn was considered by some insurer to be a
13 preexisting condition.

14 It's happened for many, many years and we now have
15 the tools and the leverage to make that stop happening under
16 the Affordable Care Act.

17 Next, please.

18 (Slide.)

19 The Patient's Bill of Rights was put into effect
20 back in September of last year. So there are a wide array of
21 things about, starting with children, the denial of --

1 prohibiting them from calling them preexisting conditions,
2 the ban on lifetime dollar limits, restriction of annual
3 dollar limits, the preventive services coverage with no
4 copays or deductibles, and then I mentioned the access to
5 children to 26; the choice of a primary care pediatrician
6 provider without referral; out of network emergency care
7 without prior authorization; improved appeals processes; and
8 prohibitions of coverage being -- losing their coverage based
9 on a mistake in their application -- all of those things are
10 now law and all of those things should be of value to some of
11 the families that the committee cares about.

12 The next slide --

13 (Slide.)

14 -- shows just a little bit, and this one was
15 developed by Christine Brown at the National PKU Alliance,
16 about some of what it means to be in these high-risk pools.
17 I'm not going to spend a lot of time on this slide, but I
18 wanted you to have her analysis of where we are in this first
19 round.

20 You see that there will be variations depending on
21 exactly how states elect to do this. Some of the states are

1 going to do it on their own. Others are going to come into a
2 federal system. There will be, I think, effectively a floor
3 in this, however.

4 Next, please.

5 (Slide.)

6 Thinking about choice and empowerment, all of
7 these protections, like guaranteed issue and portability, as
8 well as the informed choices -- many of you may have heard
9 people talking about the health insurance exchanges or the
10 exchanges, and one of the main functions of the exchanges is
11 to give people easy-to-understand, more standardized
12 information about the plans that may be available to them.

13 Next, please.

14 (Slide.)

15 In terms of coverage and benefits, I'm going to
16 say that there are protections around preventive services. I
17 think there will be a lot of discussion of this. Right now,
18 for new plans coming in, no cost-sharing is allowed for
19 preventive services rated A or B by the U.S. Preventive
20 Services Task Force.

21 There are examples, particularly PKU, in newborn

1 screening. I think one of the roles of the Secretary's
2 Advisory Committee is to be sure that that PKU screening item
3 under the Preventive Services Task Force is considered an
4 example and not that only PKU screening would be considered
5 effective. I think there is some interpretation there to be
6 done by the Secretary and I think it may need your attention
7 and reinforcement.

8 (Slide.)

9 The next issue I want to talk about really is this
10 issue of establishing an essential benefits package. HHS has
11 until the beginning of 2014 to get that work done, but
12 clearly it's already starting.

13 Next slide, please. We'll say more about that in
14 the benefits in a moment.

15 (Slide.)

16 These consumer protections really in terms of GINA
17 and the Affordable Care Act are governing certain elements of
18 health insurance. The two do not amend one another. So the
19 Affordable Care Act didn't make -- it isn't viewed
20 statutorily as an amendment to GINA, nor does it reference
21 GINA's requirements.

1 (Slide.)

2 I shared with you -- in your briefing package you
3 have an analysis by the Congressional Research Office about
4 how the two laws interact. What they found in a fairly
5 detailed legal analysis is that the GINA privacy protections
6 are stronger because GINA is civil rights legislation; that
7 the Affordable Care Act, the guaranteed issue and renewal
8 provisions do take us a little farther into protections
9 around health insurance more specifically; that there are
10 variations, just for example the GINA plans can't adjust
11 premiums or cost-sharing based on genetic information and the
12 Affordable Care Act protections apply really to premium
13 rates; then finally, that the Affordable Care Act protections
14 apply to health insurance issuers, as they are known in the
15 law, which is group and individual plans, and GINA prohibits
16 discrimination by all plans, which would include the self-
17 insured group plans. I know there's been a lot of discussion
18 among policy leaders in the field of genetics and newborn
19 screening.

20 I think this Congressional Research Office
21 analysis is very helpful in clarifying some of those points,

1 because it felt to me like there was a little bit of kind of
2 rumors starting around what the law actually said or how they
3 actually interacted. So I would urge you to take a careful
4 look at that.

5 Next slide, please.

6 (Slide.)

7 Actually, I'm going to skip this slide for this
8 time, but just to say that equal access to genetics tests was
9 not addressed in the Affordable Care Act. So next slide,
10 please.

11 (Slide.)

12 You should be on a slide about coverage and
13 benefits.

14 The benefits in this package as defined in the law
15 cover a broad set of categories. This set of categories
16 looks a lot like, it's not identical to but it looks a lot
17 like what's covered under Medicaid and Medicaid categories.
18 As most of you know, Medicaid covers many things that are not
19 on this list. So I think you have to place this kind of list
20 into that framework, that it was not envisioned that the law
21 as written, nor most people would think should it have been,

1 that all of the details of the benefits would be there, but
2 that you would set out broad categories and under that
3 specific benefits would evolve and be categorized into these
4 things.

5 Next slide, please.

6 (Slide.)

7 What that leaves us with is something that Kate
8 Pickert really has talked about as an essential benefits
9 conundrum. I thought as a reporter she did a really good job
10 in Time Magazine of talking about this, and just saying that
11 beginning in 2014 the Affordable Care Act will fine Americans
12 who don't have insurance that provides essential benefits.
13 So the idea is to eliminate Swiss cheese coverage and ensure
14 that every American has comprehensive insurance.

15 So just to pause for a moment. That's a very good
16 way to capture what that essential benefits structure was
17 intended to do.

18 Then she goes on to say: As a result, medical
19 special interest groups are already lobbying hard to get
20 their products and procedures included in the essential
21 benefits package broadly outlined in the law, but which will

1 be specifically defined. That may be, I would argue,
2 the wrong approach.

3 So let's go to the next one.

4 (Slide.)

5 And just to say that there are a lot of things
6 that will fit under those broad categories, and there is --
7 you find it in a lot of what people have written and
8 published around genetic services, this, I call it a bundle.
9 Others have called it that, the screening tests, diagnostic
10 and predictive tests in high-risk populations, genetic
11 counseling, PLUS treatment.

12 The big question to you is, where do these things
13 fit into the broad categories of service covered, and how
14 again this U.S. Preventive Services Task Force issue is going
15 to play out. I think there is a lot of room for
16 conversation, for advocacy, for the development of a refined
17 set of benefits under those categories as they exist. But I
18 think it's going to take a lot of activity on the part of
19 people who are knowledgeable about these issues, and I
20 certainly think that there is a role for the Secretary's
21 Advisory Committee in talking about this and speaking,

1 communicating directly to the Secretary about the importance
2 of this.

3 Finally, and really in all of this again,
4 something that was brought up just a little while ago about
5 really in the long-term follow-up it comes to us again and
6 again: What are the pieces that should be financed through
7 public health and population-based financing mechanisms and
8 what are the things that health insurance should be covering?

9 There's been a lot of thought into this over the
10 years. I think a lot of what was written at the time of the
11 Clinton health reform proposal is very apropos here. It
12 needs to be presented thoughtfully as the development of the
13 refined package goes forward.

14 Next slide, please.

15 (Slide.)

16 We have some lessons about the benefits issues
17 from Massachusetts. There, the Massachusetts experience
18 demonstrated that not every service or treatment will be
19 defined as essential, either in the benefit package or by the
20 medical director of a specific plan; and that the real
21 devilish details are described in the medical or clinical

1 policies of the plan; and that even if treatment is essential
2 for some, it may not be essential for all. So making sure
3 that there's coverage, making sure that the detail was there,
4 but not assuming that, for example, genetics would be listed
5 at the headline of the essential benefit package.

6 Next slide, please.

7 (Slide.)

8 There are a number of things in the Affordable
9 Care Act that relate to disparities, particularly around
10 investments in primary care for medically underserved. There
11 are other mechanisms, such as the work about community health
12 workers and investment in research about disparities. Here
13 again, I think particularly in terms of the investments in
14 research about disparities, there are some opportunities to
15 think about what is the context for understanding disparities
16 as they relate to race, gender ethnicity, and heritable
17 disease status. I know that there are
18 concerns about that under today's system.

19 Next, please.

20 (Slide.)

21 The integration with public health is I think

1 really another fundamental area here of concern. There is a
2 prevention and public health fund. It builds from \$500
3 million last fiscal year over to \$2 billion by 2015 and
4 thereafter. So there are obvious questions, like whether or
5 not funds will be committed to genetics and newborn screening
6 and whether child health and not just adult chronic
7 conditions would be a priority for this spending -- again, to
8 me an area for advocacy and communication in the public
9 policy arena about the importance of including children and
10 some of the priorities of this funding and also the important
11 role of public health to newborn screening in particular.

12 There again will be a national prevention strategy
13 and to have making sure that genetics and newborn screening
14 is on the radar in that large strategy and plan I think is
15 something that leaders in the field can help to assure.

16 Then community transformation grants seem to me to
17 be another opportunity, where state and local agencies and
18 community-based organizations are going to be thinking about
19 evidence-based community preventive health activities, and
20 again thinking about whether or not there is a project
21 related to long-term follow-up and building the evidence base

1 there under the community transformation umbrella.

2 It won't happen -- actually, I think what most
3 people working on topics related to health reform are finding
4 out is that none of this will happen -- unless leaders in
5 specific fields address the issues and start talking about
6 them.

7 The next slide, please.

8 (Slide.)

9 Getting close to the end of the taxonomy, we're
10 thinking about quality and efficiency. There is an array of
11 elements with the national quality strategy, patient-centered
12 medical home pilots, patient-centered outcomes research
13 institute, the Center for Innovation at CMS, community-based
14 collaborative care networks that give a lot of emphasis to
15 pediatric care and how you build the support around that
16 patient-centered medical home, and again community health
17 teams likewise, supporting the medical home.

18 I see a lot of opportunity to think about child
19 health systems within this. Many people are starting to
20 think about this under the CHIPRA proposals, but there will
21 be more opportunities under the Affordable Care Act as long

1 as we continue to remind people that these strategies have to
2 include pediatric approaches.

3 Next, please.

4 (Slide.)

5 I would just close by sharing with you a quote
6 from Tom Murray about there being a "broad range of values
7 that we want our health care system to embody and pursue --
8 not just liberty, but also justice and fairness and
9 responsibility, and medical progress, and privacy and
10 physician integrity, among others." I think this cross-
11 section of what the Affordable Care Act has been able to
12 achieve in many ways falls short of people's expectations on
13 any one of those items, but in many ways exceeds everyone's
14 expectations for what we thought might actually get done
15 through the Affordable Care Act. The public messages don't
16 begin to scratch the surface about the actual potential to
17 improve things like justice and fairness and medical progress
18 and the whole service delivery system as well as coverage.
19 But there clearly are elements across all of those that are
20 very important to the whole population, but I think can be
21 also particularly important to those with heritable

1 disorders.

2 So let me stop there and hand it over to Brent.

3 CHAIRPERSON HOWELL: Thank you very much, Kay.

4 Why don't we go ahead with Brent's presentation
5 and then we'll have comments and questions for the both of
6 you.

7 CURRENT EFFORTS RELATED TO IMPROVED

8 FINANCING AND HEALTH CARE REFORM

9 DR. EWIG: Terrific. Can you remind me about how
10 long we want to go? Ten minutes?

11 CHAIRPERSON HOWELL: Sounds good to me.

12 DR. EWIG: Okay, I'll try to do that.

13 First of all, what a pleasure to follow Kay,
14 because she did all the hard work and the slides are just
15 terrific and really kind of lay out in a factual basis here's
16 what's in the law and the analysis of how that will impact.

17 Let me also say thanks for the invitation to spend
18 some time with you today with this prestigious body and
19 acknowledge up front the leadership that Dr. Chris Kus as a
20 voice for state public health has provided, Dr. Van Dyck and
21 Dr. Howell. I think when we look at newborn screening from

1 AMCHPs perspective it's something we hold up as a public
2 health success story. When we think of where we were a
3 decade ago to where we've come now, obviously a lot of
4 organizations and groups leading that -- the March of Dimes
5 that I see in the room and others. It's really been
6 tremendous progress. So the first message to you all is
7 thanks for your leadership and your work to help make this a
8 success story.

9 I also want to acknowledge Lauren Raskin Ramos,
10 who's AMCHP Director of Programs, is in the audience, has
11 been engaged with these issues for a long time and brings
12 expertise.

13 Again, it's a real pleasure to follow Kay because
14 she's so smart and laid these out so well that it kind of
15 makes it an easier job for me to just kind of provide a few
16 reactions, a little bit of perspective. Where I wanted to
17 focus is really on the implementation part of this. To state
18 the obvious, I can't really talk about implementation without
19 talking about the politics of health reform right now.

20 I keep trying this line and it falls flat, but I
21 try to quote that great line from Casablanca where he says

1 "I'm shocked" -- shocked to see that people are playing
2 politics with health care in this country. If you heard the
3 President the other night, he said he's heard a few rumors
4 that there's some disagreement still about this law. But I
5 think he really tried to set the tone on moving forward.

6 The bottom line message as a representative of the
7 state MCH programs that I want to bring to you here is that
8 we are well aware of the politics around this and how that
9 intersects with policy and affects the varying levels of
10 enthusiasm, but at the end of the day we are Executive Branch
11 employees and in the American system of government our job is
12 to implement and execute the law. We let the legislating to
13 the legislators.

14 So again, with this body charged by the Secretary
15 as an Executive Branch official, I think we kind of hold that
16 focus, that we need to be aware of everything that's in the
17 air and swirling around and how that affects the various
18 opportunities that are before us in this law, but at the end
19 of the day that's our job, to move forward in implementing as
20 best we can, maximizing these opportunities, and keep doing
21 it until somebody says stop.

1 Obviously, to pick up on that -- actually, I want
2 to step back real quick, because I always heard that when you
3 do a public presentation that if you share something personal
4 with the group that that will give you -- they'll remember
5 you more if you do that, no matter what you say. So what I
6 wanted to say is I come to you here today both as a public
7 health professional, but also as a father.

8 I have two daughters, one that's two and a half
9 and one that's just eight weeks old. So this morning as I
10 was getting ready with the eight-week-old, I said: Remember
11 when you were in the hospital and you had the heel prick and
12 you gave that blood sample? She's very advanced and she
13 said: Yes, I do; that was kind of traumatic.

14 I said: Well, I'm going to speak to those people
15 today that have led that system. And she said: Oh; well,
16 good luck. So really the professional and personal parts of
17 my life have intertwined with raising two young daughters.

18 CHAIRPERSON HOWELL: What we would like to know,
19 Brent, does your wife know that she had newborn screening?

20 DR. EWIG: We talked about it right in the
21 hospital, yes. I was very pleased to see that.

1 CHAIRPERSON HOWELL: Excellent.

2 DR. EWIG: So here's the things that we've been
3 tracking and the kind of help that we've been trying to
4 provide to the state MCH programs. Obviously, a lot of what
5 we do is report on what's happening here in Washington so
6 they understand how that's playing out. It's fair to say the
7 last months since passage of this law have been quite an
8 adventure. I think a lot of initial enthusiasm and then by
9 the fall it was really clear that the number of challenges
10 that were being erected, both at the federal level and the
11 state level, and seeing how as lawsuits were filed and how
12 people were interpreting and things were going, and obviously
13 the elections really setting a different change in tone.

14 So following that through, obviously one of the
15 headline-grabbers has been the efforts to repeal the law. I
16 think again why I mention that is because it creates this
17 confusion of, okay, so we know what's in the law, but is it
18 going to stand and how does that play out?

19 So you've all seen the House of Representatives
20 has already had a vote on repeal, voted, largely on party
21 line votes or a party line vote, to repeal, and that's

1 passed. The Senate's made it very clear that they're not
2 going to take that up, and the President's made it clear that
3 he would not sign any legislation that significantly
4 undermines one of his top priorities.

5 So what we've taken away from that and have been
6 sharing far and wide is essentially that I think it's safe to
7 say that the federal statutory framework, all the things that
8 Kay talked about, at least for the next two years are
9 relatively safe.

10 The second tier of that is stated efforts to go
11 after the funding, and that's where the prevention fund and
12 the link to the potential support for newborn screening
13 activities really comes into play, because we've already seen
14 one attempt to use the prevention fund, which started at \$500
15 billion last year, \$750 million in this year -- did I say
16 "billion"? Sorry. \$500 million in the first year, \$750
17 million in this current fiscal year.

18 In the next fiscal year that starts October 1, a
19 billion dollars of new money will become available to support
20 broadly, the way the law reads, any program authorized under
21 the Public Health Service Act. So Newborn Screening Saves

1 Lives Act activities obviously would fall under that and
2 would be potentially supported by that fund.

3 What we've seen is attempts to use that fund as
4 offsets for other policies. There has been a vote on it in
5 the Senate once that failed to do that, but I think it's fair
6 to say that in such fiscally tight times any money that's out
7 there that hasn't yet been obligated is going to be
8 vulnerable. So that's something we track very closely, with
9 literally dozens of other groups in the public health
10 community, to assure that those investments made in the
11 Affordable Care Act remain strong and that people understand
12 why those are critical investments.

13 So that's sort of the second tier of action on the
14 de-funding. The third tier that we've been watching and I
15 think is important to watch is the litigation around the law.
16 Everyone's seen a number of states that have joined in
17 lawsuits that have really been challenging two things: one,
18 the individual mandate, and that gives people an opportunity
19 to talk about it. You can't eliminate preexisting conditions
20 without having the individual mandate, so the repeal and
21 replace dialogue there becomes interesting then for

1 policymakers that say, we want to preserve the preexisting
2 condition exclusion but take away the individual mandate. It
3 makes it essentially you're saying you can have a car
4 accident and then buy insurance. So it doesn't quite work in
5 an insurance world.

6 It'll be interesting to see how that plays out. I
7 think of the rulings already, you've seen that two judges
8 have ruled that the bill is constitutional, one that it's
9 not. I think you fast-forward there, everybody has kind of
10 from the beginning understood this will go to the Supreme
11 Court, where they'll have the final say. I think from our
12 perspective and what states are watching is they want to see
13 that happen in a timely way so that it can be litigated, be
14 resolved, and at the end of the day they'll have the final
15 word and we'll know that federal statutory framework, how
16 that will go forward.

17 The second lawsuit -- and I think what we had
18 heard is that a decision on this is out of the Florida court.
19 That lawsuit has a different wrinkle in that it's a state
20 challenging the Medicaid expansion as well as the individual
21 mandate. Obviously, for children's coverage and the role

1 that Medicaid plays is a primary for so many, for nearly 40
2 percent of births, obviously the future of that program is of
3 great interest and seeing how that case is resolved will be
4 of great interest.

5 As I mentioned, I think we're expecting a ruling
6 on that shortly, and we again expect that it will be
7 consolidated with other cases and run up to the Supreme Court
8 and they'll have the final say.

9 So that's kind of the big picture of tracking
10 what's happening with the politics of health reform. To
11 follow up on Kay's excellent framework, I think what I just
12 wanted to share with you is the way we at AMCHP have
13 approached this is really breaking it down, because the bill
14 is so large and complex, and everyone understands that, and
15 there are so many different moving pieces here. We've tried
16 to say, if everything is a priority nothing will get done.

17 So with our membership of state MCH leaders we've
18 tried to really break it into a couple different slices. One
19 is that we've said let's look at the coverage provisions, the
20 benefits provisions, and then the public health and public
21 health system prevention investments, so those three buckets,

1 and then let's look at what are the immediate opportunities
2 and how we have kind of defined that is where is there money
3 available, the mandatory appropriations that were made that
4 began in fiscal year '10, what are the intermediate
5 activities of the policy-setting that happens now, and what's
6 the more long-term focus on really looking at those 2014
7 deadlines when the expansion really kicks in.

8 Along those lines, and again to preserve as much
9 time I'll be brief here, and we can come back to it. But on
10 coverage, as Kay said, I think the step forward that the
11 Affordable Care Act makes both on the affordability but the
12 stability and coverage, that with that individual mandate
13 what we're saying is people cycling on and off, that those
14 days would be over. I think from a state perspective this
15 idea of Medicaid eligibility, where currently systems have
16 been linked to the pregnancy status of a woman, that she's
17 eligible when she's pregnant, but then she loses it in
18 between pregnancies; it's obviously hard to do pre-conception
19 and inter-conception care when women's coverage is tied to
20 that status.

21 So the potential to stabilize those markets for

1 women and children we think is a great step forward.

2 The other piece there that I think the states are
3 beginning to think through is that culture of enrollment,
4 that now, as you know, many states, largely driven by budget
5 considerations, make it hard to become eligible and to
6 maintain that eligibility. An individual mandate saying you
7 have to cover this changes that whole perspective. Rather
8 than keeping people off the rolls, the real challenge for
9 states is how do they design integrated systems between their
10 exchange and Medicaid that assure that there's no wrong door,
11 that wherever you apply we find wherever you're eligible and
12 get you enrolled in stable, affordable coverage.

13 So I think that will be a revolution. I think
14 there's a lot of best practices out there, whether they're
15 called no wrong door or express lane eligibility. There's a
16 lot of things states are looking at and sharing and building,
17 and our colleagues at the Governors Association and Medicaid
18 directors, insurance commissioners, and the National Academy
19 for State Health Policy have really been leading a lot of
20 that work to lay the groundwork.

21 Benefits again for this group I think is where

1 there's a lot of opportunity and focus. Our bottom line
2 message at AMCHP all along has been we want to see as many
3 people covered as possible, and we want a benefits package
4 that is as comprehensive as possible to meet the unique needs
5 of women and children, particularly children with special
6 health care needs.

7 Our message again on that has always been: If we
8 can design a system that works well for children with special
9 health care needs, then it will work well for all children.
10 That's a line I stole from Dr. Kus, so I do, so it's not
11 plagiarism, the air quotes.

12 But that's really been our message. In looking at
13 the benefit design, I think we were very pleased with seeing
14 the original prevention reg that put into place the statutory
15 framework around Bright Futures for Children, being the
16 standard of prevention services care with no cost-sharing for
17 children. That reg is out.

18 I did want to call people's attention, because it
19 says -- as you all know, the way the benefits were set up,
20 that anything that receives an A or a B from the Prevention
21 Services Task Force will be covered with no cost-sharing.

1 Again, this is in new plans, so there is that issue of
2 grandfathering. But for new plans, and largely that means
3 people that will be gaining coverage through the exchanges,
4 that's the standard, the beginning of the standard.

5 Then for kids the standard is: and anything
6 that's recommended by "Bright Futures." So they essentially
7 set the periodicity schedule of "Bright Futures" becomes the
8 standard. Then it's really important: They then added on
9 and then said: And the standard panel recommended by this
10 group for newborn screening shall be covered with no cost-
11 sharing.

12 So I think it's in there. It's in the HHS
13 prevention reg and that provides us a solid foundation to
14 move forward. But as you all know, that's just for the
15 screening part, and it's the long-term care follow-up that we
16 want to make sure the benefits package is adequate to cover
17 that.

18 Where that stands right now, as Kay laid out, the
19 law did the outline: Here's the services that should be
20 covered. Pediatric services are listed. Obviously a lot of
21 room for defining that. So the Department has recently

1 contracted with the Institute of Medicine to begin that
2 process. They've put together a panel that's met just a few
3 weeks ago, to begin that dialogue of what's the scientific
4 basis and process that we should look at to set that
5 essential benefits package. But, knowing at the end of the
6 day it will be HHS that defines that, there will be plenty of
7 opportunity for opportunity comment, and that's where I think
8 groups like yours and AMCHP and others and all of us in the
9 MCH community will be watching carefully and using that
10 opportunity to say, here's a package that we think is
11 adequate for children with special health care needs
12 obtaining the genetic services and follow-up that will be
13 needed.

14 So that's where things stand on benefits and where
15 our advocacy role has been. On the prevention health and
16 public health system improvements, these are twofold, where
17 we talk about the investments and promoting the medical home
18 model. I call everyone's attention, the CMS Innovation
19 Center that's created by the law and has a mandatory \$10
20 billion appropriation over ten years, so essentially some
21 real resources there, that's going to be the centerpiece of

1 the Department's efforts to really promote medical home,
2 promote that experimentation with bundling of payment
3 services to drive more integrated care.a

4 They have issued a letter. There's a new acting
5 director. Dr. Richard Gillfillan, I believe, has been named
6 the acting director of that center. I think that's the place
7 to watch what they put out. In their initial letter, they
8 put out guidance to state Medicaid directors on how to access
9 the money that's available for the medical home pilots. I
10 think that's going to really be a driver of innovation in
11 medical home and integrated care. That's where I think we
12 can focus and make sure we're connecting with those folks.

13 Then that's where we look at the prevention fund.
14 I touched on that up front, that we see this as a huge step
15 forward, a recognition in federal law that public health has
16 been chronically underfunded, needs a stable mandatory public
17 health fund that grows up to be \$2 billion a year annually in
18 its fifth year. That's where we and other groups have said
19 we understand that a lot of the focus early on is going to be
20 on chronic disease and to the extent that that's focused on
21 promoting good health in women of reproductive age, that will

1 improve birth outcomes, but as the fund grows and resources
2 become available we want to make sure that an adequate
3 portion is going specifically to maternal and child health
4 programs, so something like the Newborn Screening Saves Lives
5 Act, which you've all seen how hard it is to get that fully
6 funded, that's the opportunity.

7 I think, to state the obvious, in an environment
8 like this, where there's so much pressure on the budget, on
9 discretionary, having that pot of mandatory funding. to make
10 sure we protect it and to make sure that that's deployed
11 adequately to MCH programs is going to be both a huge
12 challenge and an opportunity in the coming years.

13 Then finally, I'd leave with this thought on
14 budget issues. We are obviously facing unprecedented
15 challenges, and you arrived in town when the announcement was
16 that we're at yet another record high deficit. The level of
17 cuts that are being discussed on Capitol Hill I think we have
18 a great deal of concern are just going to be crippling to a
19 number of state MCH program efforts.

20 I know there's specific line item funding in the
21 Newborn Screening Saves Lives Act that goes to patients. I

1 think it's in the neighborhood of \$5 million. The MCH block
2 grant in Title V, that has about 500 times more money that's
3 going to states, obviously not all for newborn screening, but
4 to promote systems serving all women and children. The level
5 of cuts that we're looking at being proposed for those
6 programs are truly alarming. So we are obviously paying a
7 lot of attention to that and the potential that that really
8 would undermine a lot of the work that you have helped head
9 and the progress we've made.

10 So that's part of our message there, and it's a
11 bottom line message that we understand the need for deficit
12 reduction, but cuts in this area are counterproductive,
13 because if we're not catching things early, if we're not
14 preventing what we know how to prevent, we're just simply
15 going to pay more later. So it's a real tough argument,
16 though, to make in this environment.

17 But I don't want to end on a downer note. I just
18 wanted to make everyone aware of the critical nature of the
19 situation, and then turn and have some dialogue with you.

20 CHAIRPERSON HOWELL: Thank you very much, Brent.

21 Are there questions or comments for Kay or Brent

1 about their excellent presentation?

2 (No response.)

3 You obviously answered all the questions.

4 Well, tell your eight-week-old when you're having
5 a conversation tonight that we wish her well and we hope her
6 screens were all normal, with no false positives.

7 DR. EWIG: She failed -- my first daughter failed
8 her newborn hearing screening. But on the retest she turned
9 out fine. So we had an experience with that, of what does
10 that mean. But everything else has been great.

11 CHAIRPERSON HOWELL: Excellent.

12 DR. EWIG: Thank you all for the chance to talk
13 with you all, and again the leadership that you've been
14 providing has been really appreciated.

15 CHAIRPERSON HOWELL: Well, thanks very much for
16 all your work that you're doing.

17 Ms. Harris has the ability of being very brief and
18 succinct, and so I'm going to ask Alaina if she'd be good
19 enough to move her presentation up to before lunch, and I
20 think we can still get to lunch on time. She is going to --
21 Ms. Harris you all know, I think. She is a Public Health

1 Analyst in the Genetic Services Branch. Today she's going to
2 discuss the committee's options for the report to Congress,
3 which is due in April. That report, as you know, is mandated
4 in the law that oversees this committee.

5 **THE COMMITTEE'S ANNUAL REPORT TO CONGRESS**

6 ALAINA HARRIS, M.S.W., M.P.H.

7 MS. HARRIS: Thank you, Dr. Howell.

8 (Slide.)

9 I don't have a personal story with newborn
10 screening to share to make myself memorable. I have a dog,
11 but she was not screened at birth.

12 CHAIRPERSON HOWELL: That's why she's barking so
13 much.

14 (Laughter.)

15 MS. HARRIS: I will say that Annabelle did not
16 talk to me this morning, so I have no good wishes.

17 CHAIRPERSON HOWELL: I think that dog's requesting
18 screening be done immediately.

19 MS. HARRIS: Realizing that I'm keeping you guys
20 from lunch, I will keep this brief.

21 As Dr. Howell has mentioned, for our annual

1 report, the committee's annual report to Congress, this is
2 something that is required in the Newborn Screening Saves
3 Lives Act legislation. So for today we'll briefly go over
4 the legislative requirements. I want to present the report
5 outline that we have come up with. And then I'd like to get
6 some feedback from the committee on that outline, and then
7 also some possible current and future issues that we want to
8 include other than funding in this report to Congress.

9 (Slide.)

10 Section 1111 of the Newborn Screening Saves Lives
11 Act reauthorized and expanded the activities of our
12 committee. Today we're going to talk about part (e), the
13 annual report.

14 (Slide.)

15 In there, the legislation required three years
16 after the enactment of the legislation that we would publish
17 a report on peer-reviewed newborn screening guidelines,
18 including follow-up and treatment.

19 (Slide.)

20 The legislative intent was to provide a summary of
21 the accomplishments to date and future plans. This goal of

1 the annual report, if you tune me out for the rest of this,
2 please just listen to this for our discussion. For us to
3 really have this opportunity, we're going to have Congress's
4 eyes for a bit with this report, and one of the things is
5 using this to outline some next steps for Congress in terms
6 of supporting the work of our committee and supporting state
7 newborn screening programs.

8 (Slide.)

9 Here is the outline that we have come up with so
10 far. We're starting with section -- well, we start with the
11 background on newborn screening and screening for children
12 with heritable disorders; go over section 1111, which is
13 providing a legislation summary, and then an overview of the
14 committee, the subcommittees, workgroups, etcetera.

15 We will discuss the committee's activities in the
16 previous three years, starting with a discussion of the
17 recommended uniform screening panel. That's the RUSP. So
18 we'd at the conditions that have been examined and the
19 committee's decision processes out of Dr. Perrin's group.
20 We'd also look at the committee's model decision matrix and
21 evidence review process. We'd follow up with a discussion of

1 other committee recommendations and then committee reports
2 and briefing papers.

3 Our next section would be a state of the states
4 capacity to screen for the recommended uniform screening
5 panel. This includes information on the heritable conditions
6 that states require, incidence and prevalence of these
7 conditions, and then the other items related to section
8 1111(b)(6). In your briefing book I did include the
9 legislation, and there are about 14 different things that are
10 part of this section 1111. I didn't list them here. You are
11 able to read, I know, so if that is of utmost importance to
12 you you can find that information there.

13 (Slide.)

14 Then we thought it would be nice to follow up with
15 part of the report on the implementation of the RUSP to the
16 heritable disorders program, so the rest of the Newborn
17 Screening Saves Lives Act. Section 1109 is grant programs
18 for system infrastructure, so that's out of HRSA. This will
19 be a report on the regional collaboratives, the effective
20 follow-up on newborn screening, those different grant
21 activities.

1 Section 1110 is the grant programs to evaluate the
2 effectiveness of screening, counseling, and health care
3 services. This is something else that also comes out of
4 HRSA, early and continuous screening in the medical home.

5 Section 1112 was the Clearinghouse for Newborn
6 Screening Information, which we heard about from Dr. Trotter
7 today, would be covered in that.

8 Section 1113 is the laboratory quality assurance
9 program. That's with CDC and Carla Cuthbert's group, so we
10 have a report from them.

11 The Inter-Agency Coordinating Committee out of
12 section 1114, when that happens. Section 1115 is
13 contingency planning with CDC and HRSA. Then 1116 is the
14 Hunter Kelly Research Program out of NIH and NICHD.

15 We'd follow this up with current issues, future
16 issues, and then appendices of articles and briefing papers.

17 (Slide.)

18 So any discussion on the outline as you see it as
19 presented?

20 (No response.)

21 You all are hungry.

1 DR. KUS: One comment. Could you go back to the
2 slide where it says "State Capacity."

3 (Slide.)

4 You know, one of those things that bothers me when
5 we have this discussion, it says: "The states' capacity to
6 screen," as opposed to "The states' capacity to ensure
7 screening, diagnosis, and treatment." I think that's a point
8 we have to drive home to folks, that it's not just the test;
9 it requires lots of work and resources afterwards.

10 DR. VOCKLEY: I wasn't quite sure where to start
11 making this comment, but maybe I'll make it here because I
12 think it follows up Chris'. That is, we do here have to make
13 these recommendations or we try to make recommendations, but
14 we're dealing with what we see as the optimum. Our goals
15 here are to say this is what's good for kids.

16 When you go into the states, they're really now --
17 if this is the ivory tower, they're bricks and mortar.
18 They're trying to implement our recommendations. I have an
19 interesting conundrum because I'm on this committee, I'm the
20 chair of our state newborn screening technical advisory
21 committee, plus I'm the director of a newborn screening

1 follow-up clinic. If the states are brick and mortar, at the
2 local level we're out in the field with no protection. We're
3 working to now take care of all these kids that the states do
4 manage to plan for.

5 So this is a continuum of obligation that starts
6 with this committee. I'm not exactly sure where we -- how we
7 need to recognize this, but somebody sure does, because I'll
8 tell you, out in the field it's raining, or snowing in this
9 environment. But we're getting bombed and it's really
10 difficult to see how we're going to implement everything that
11 comes out of the committee.

12 It is funding, yes. There's no denying that. But
13 just the whole process of making sure that we're closing the
14 loop here I think is very important.

15 CHAIRPERSON HOWELL: I think that most are very
16 much aware of the things that Gerry points out.

17 Alan.

18 DR. FLEISCHMAN: Just an additional comment. I
19 think this is an extraordinarily successful federal advisory
20 committee. It is extraordinarily productive in the last
21 several years. I think it has a tremendous future agenda.

1 So that in that section on the future, the implementation of
2 the Secretary's recommendation around SCID is really quite
3 astounding. We've seen a lot of tremendous work there and
4 the states are really beginning to give us the data to
5 support that.

6 The work that's already gone on in thinking
7 through the critical cyanotic congenital heart disease issue
8 is extraordinary and we ought to note that and note that it's
9 moving forward. The report that Tracy gave on the
10 educational planning, which is part of the bill as well, on
11 this committee's job, is extraordinary.

12 So there's a lot of positive future look for
13 Congress at what will come continuing, as we continue, and
14 the extraordinary success. So I think you've tracked the
15 success and you should be very proud of the future planning
16 that's going to go on in the next three years, and we're
17 going to have just as much success.

18 MS. HARRIS: I do want to follow up -- thank you,
19 Alan -- with Dr. Vockley. Some of those section 1111(b)(6)'s
20 follow-up, implementation, etcetera -- and I thank you for
21 bringing up the reminder that it's not just screening;

1 there's a whole component to screening, short-term, long-term
2 follow-up, etcetera.

3 CHAIRPERSON HOWELL: And I'm sure that will be
4 certainly amplified.

5 I think that, to follow up on what Alan has said,
6 it's been remarkable to see the states move rapidly ahead in
7 the area of newborn screening, although all these
8 vicissitudes we're aware of. The other thing is that most of
9 the patients that we're identifying have always been there.
10 In other words, they've not been formally recognized. They
11 have badly been treated under some circumstances. We really
12 aren't finding new patients. We're simply identifying them
13 earlier. So I think that that's the other thing, that we are
14 not inventing patients; we're discovering them and hopefully
15 benefiting them.

16 But I think you can make a positive thing. I
17 think the NIH, Tina and her group, can certainly put together
18 some impressive data about research, some of which we've
19 heard about here for the Hunter Kelly program. The CDC
20 obviously has lots of things to put in. So I think it should
21 be a very strong report.

1 Are you planning to circulate this to the
2 committee? It needs to go in soon.

3 MS. HARRIS: It does need to go in soon. Our next
4 steps, we will contract with Elissa Johnson to help do the
5 pulling together of the report. Different folks have been
6 approached with writing different aspects of the report, but
7 Elissa would pull that together. We'd like to get that to
8 you, a draft, some time in the beginning of March.

9 DR. LLOYD-PURYEAR: To date, only NIH has
10 responded with their piece. So we're waiting for the other
11 federal agencies. But we're also assigning several parts to
12 our various cooperative agreements.

13 CHAIRPERSON HOWELL: Congratulations to the NIH
14 hardworking people that got their thing together. You should
15 use that as an example, Coleen.

16 (Laughter.)

17 MS. HARRIS: I do want to follow up, though, on
18 what Dr. Howell said and Dr. Fleischman. One idea I had was,
19 in terms of a way to maybe liven up this report, is if we
20 could maybe have some different text box sections of people
21 that have been positively affected by newborn screening

1 interspersed throughout the report. We have not had a chance
2 to talk with the Genetic Alliance yet about their consumer
3 task force, but I know, having been on their calls, that they
4 are working on also coming up with positive stories, and that
5 might be a way to showcase what we're hearing from our
6 consumers.

7 CHAIRPERSON HOWELL: The regional collaboratives
8 could obviously provide you with persons that have been
9 identified that would be suitable during this time frame.

10 MS. HARRIS: Of course.

11 CHAIRPERSON HOWELL: And ask them if they'd be
12 willing to be interviewed. Certainly there clearly are
13 patients, children, that have benefited.

14 Are there other comments or questions?

15 (No response.)

16 Thank you.

17 DR. LLOYD-PURYEAR: We're not quite done.

18 MS. HARRIS: We did do a little brainstorming in
19 our office as well on some possible current issues that we
20 might want to touch on -- obviously, privacy and
21 confidentiality issues, public perception, and then the vast

1 work that's coming out of the Health Information Exchange.

2 I also heard, in terms of current issues,
3 highlighting the implementation of SCID, CCCHD, and the
4 newborn screening awareness campaign.

5 Anything else?

6 (No response.)

7 Then in terms of possible future issues, point of
8 care testing, timing of screening beyond the newborn time
9 frame. As has been noted today, we are tasked with newborn
10 and child screening. Infrastructure needs as we move to this
11 point of care testing and screening; integration of screening
12 for heritable disorders across the lifespan.

13 Anything else or any comments about these topics?

14 CHAIRPERSON HOWELL: Are you going to comment on
15 anything about carrier screening, which we've had a good bit
16 of recent activity on?

17 MS. HARRIS: Yes.

18 CHAIRPERSON HOWELL: Okay.

19 MS. HARRIS: I don't know what we're going to
20 comment on it, but yes, we will.

21 (Slide.)

1 Well, here is my contact information and I
2 appreciate if there are any other thoughts and comments that
3 people want to make, please feel free to send me an email or
4 give me a call.

5 Thank you.

6 CHAIRPERSON HOWELL: Thank you very much, Alaina,
7 for that very succinct presentation.

8 We are slightly ahead of schedule, and so we'll
9 stop now for lunch, and we will return promptly at 1:00
10 o'clock so that we can again try to go through the important
11 activities this afternoon and still get out actually a little
12 early.

13 Thank you very much.

14 (Whereupon, at 12:00 o'clock noon, the committee
15 recessed, to reconvene the same day.)

16

17

18