NewSTEPs
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January 2014 Short Term Follow Up Webinar

January 6, 2014

Presentations:

- State Profile Vermont—Cynthia Ingham, RN, BSN
- The Importance of Identifying False Negatives—Marci Sontag, PhD
- Using a Secure Portal (Share Point) for Communications and Data Collection—Brigitte Dufour

Moderators:

- Thalia Wood, MPH, Specialist, NewSTEPs
- John Thompson, PhD, MPH, MPA, Co-chair of the Short Term Follow Up Work Group

Please direct all comments/questions pertaining to this presentation to Thalia Wood at Thalia.wood@aphl.org or 240-485-2701.

Thalia Wood: Okay John if you want to go ahead and start by introducing the call.

John: Hello everyone and welcome to the second short term follow up webinar sponsored by NewSTEPs at the APHL.

Mark: Hi [inaudible 00:00:50].

John: Very glad that you've joined us today, and give you just a brief overview of what we'll be hearing about and then we'll press forward. Our state spotlight today is Cindy Ingram from Vermont will be sharing some words about their program, excited to hear from her. The main portion of our discussion today will be about false negatives in newborn screening. We'll have a brief introduction from Marci Sontag out of Colorado School of Public Health and then Brigitte Dufour from the Arizona Newborn Screening Program has a presentation also for us.

Then we will have a chance for questions and answers and then at the tail end of our seminar today, Marci is going to speak again, this time she is going to speak on the NewSTEP's response to the Milwaukee Sentinel

Journal articles that have come out recently, so we will proceed as planned. Thank you all for joining us.

Thalia Wood: Thank you John. Marci let me remind you to go ahead and mute the lines for all but the presenters then at this time.

Telephone: The conference call has been muted.

Cindy Ingham: Thalia?

Telephone: The conference has been unmuted.

Mark: You muted yourself.

Thalia Wood: I thought the presenters would not be muted, so Cindy you'll have to do a star 7 and unmute yourself to talk.

Cindy Ingham: Hi, this is Cindy, can you hear me?

Mark: Yes.

Thalia Wood: Yes.

Cindy Ingham: Okay then I'll just go right ahead. Hello everybody and Happy New Year from Vermont, it's just the second least populous state of the United States. [Inaudible 00:02:58]. We're about 95% white. We have under 6,000 births a year. Also the Americas Health Ranking bumped us form number one to number two after Hawaii as the healthiest state. That gives you a picture of where I live and work.

Next slide, this is a little hello postcard from Burlington [inaudible 00:03:31] city, and by large, I mean about 42,000 people.

Thalia Wood: Excuse me for one second please. Since we are not able to mute everyone on the call, could we ask people to please mute your phones so that there is not a lot of background noise, thank you.

Cindy Ingham: Yeah, we are getting some feedback. Okay, so that red arrow you see points to the Department of Health where I work. Mountains in the background and we are 45 miles south of the Canadian border. If you haven't been here you should be. As far as the staff for the newborn screening program, I am the only full-time person here and I do all

aspects of the program from quality assurance to billing, grants, contracts, legislation, program planning, training, that sort of thing. I work half the time administrative assistant. I have one nurse whose cross trained to back me up if I am on vacation or out of state. It's just me and Trish, the Administrative Assistant.

I will show some statistics from 2012 just to show you what we're dealing with; about 6,700 births, 75 were eligible to have newborn screening, of them, most of them were screened in Vermont, a total of 35 screened out of state. In this state parents can refuse screening their babies for any reason at all. Each year we do have less than one-half of one percent who refuse screening. So for 2012, 99.7 percent of our eligibles were screened. Since 2003 we've been able to document 100% of all babies born in this state either screened, died before screening or were counseled and refused and signed a refusal form.

So we aren't missing any that I know of. [Inaudible 00:05:57] against the birth certificates that have been filed. There are always a handful of babies whose births aren't registered right away, the midwives usually let us know. In 2012 with the conditions that were identified, [inaudible 00:06:23] that were diagnosed as being true positives so like most of you, congenital hypothyroidism always gets the prize for the most disorders screened.

Had some homocystinuria, one CF, [inaudible 00:06:43] galactosemia. In addition, of course, we also pick up other disorders or conditions; CF carriers, sickle cell [inaudible 00:06:56] deficiency. Generally each year we'll pick up one maternal condition that is reflected [inaudible 00:07:04] and other things through and I don't have a slide for this, but so far in 2013 our preliminary diagnosed conditions picked up are eight hypothyroid babies, a baby with sickle cell disease, [inaudible 00:07:25] with cystic fibrosis, biotinidase deficiency, one with PKU, one with citral anemia, another baby with what turned out to be Pyruvate kinase deficiency, which has a triple implication, [polyman 00:07:45] deficiency and in a variety of carriers.

Even though our numbers are small of the experience of dealing with all sorts of conditions, what we're working on now as are most of you, are adding CCHD and SCID screening. CCHD is on the front burner, and actually gotten started at all hospitals in the state are at some form of screening. One of the physicians, it's only tertiary care facility and I are

working on [inaudible 00:08:31] on trainings for the academy of pediatrics, [inaudible 00:08:42] chapters here in the state so that the primary care providers are [inaudible 00:08:48]. Other things that are going [inaudible 00:08:54] has become more of a part of other public health efforts such as [inaudible 00:09:03]. I also am on the Refugee and Immigrant Health Services Network, which meets approximately every other month.

I do a lot of training with a monthly one to one orientation for pediatric residents who are starting their NICU rotation, also for pediatric fellows, periodically at pediatric grand rounds. I started teaching in the field of chemistry of all things, which was my worst subject in college, who knew. I teach a class for undergraduate students in clinical chemistry. [Inaudible 00:09:49] for the Vermont Society for Clinical Laboratory Science as well as [inaudible 00:09:57]. They're always very interested in new and newborns, how we can work together. Next week I'm going to be doing a [inaudible 00:10:08] a screening for metabolic conditions.

One thing I was thinking about that I have found very, very, essential [inaudible 00:10:29] program, periodic face to face meetings with key external staff, such as the neo-natology and NICU staff, the consultants, the metabolic, fibrosis and hematology consultants that I turn to, sometimes every day, for guidance. I don't know if Carol Johnson is on the call today, but she has sent out a call about working with endocrine [inaudible 00:11:06] those are the [inaudible 00:11:08] that are much easier [inaudible 00:11:11] person face to face when you go over your protocols together and look at types of issues that have come up [inaudible 00:11:18] relationships. So, that's a very quick overview of what's going on here in Vermont. Anyone [inaudible 00:11:27] and my contact numbers and so forth and I am happy to take some questions.

Thalia Wood: Actually, Cindy this is Thalia, I think we'll hold questions until the end. I want to remind everyone please, please mute your phones or turn down your speakers on your computer because we are getting an awful lot of feedback and it's making it very difficult to hear.

Cindy Ingham: Yes.

Thalia Wood: But we will get questions at the end, thank you.

Cindy Ingham: In that case I will mute myself now too.

Jelili: Yes, Thalia, I think, this is Jelili, let's do something else. Is Dr. Sontag the next speaker?

Thalia Wood: She is, but she is actually the chairperson today since we're having problems with our phones here, Jelili.

Jelili: Okay.

Thalia Wood: So, Marci, if you want to mute everybody and have the speakers just do star 7 to talk, that might be better.

Jelili: Exactly, I think that would appropriate, so Marci if you don't mind muting everyone and just pressing star 7 and for other speakers, when it's your turn press star 7 to speak.

Marci Sontag: I will do that. The last time I did that I lost the ability to hear everyone. If that happens, someone send me a chat and we'll try to work it out.

Jelili: Well you will lose the ability to hear everyone, but that's what I did last time and I just have everybody do star 7 to talk.

Marci Sontag: Okay.

Jelili: Then I unmuted at the end.

Telephone: The conference has been muted.

Marci Sontag: Okay, so I have muted everyone and I am going to go ahead with my presentation next. Thank you so much, everyone, for joining us, and I have been asked by John Thompson to start this off with a presentation I gave a couple of years ago about the importance of identifying false negatives. My father was a fisherman so I put the idea of newborn screening in the context of fishing.

So, as you all know, in public health newborn screening, all babies are tested or nearly all babies are tested, and we want to cast that net broadly so this gave us that fishing net to cast that out broadly to ensure we capture all babies who are at risk of having the disease, but we really want to try to balance that false positive with the sensitivity of the test so we're not calling back too many babies in for the number of babies that we might be missing, with the thought that missed cases really could have devastating outcomes.

The false negatives in newborn screening really could come from many different sources and we capture almost all babies or at least we think we do, but this is really one of the most important quality indicators of our newborn screening programs is how many babies are we capturing? So our public health newborn screening systems across the country capture most babies with the screened diseases, but we really aren't able to calculate the missed case rate, whether they be false negatives, biologic false negatives or missed for other reasons. We have trouble calculating that missed case rate because we don't have systems to collect the data.

We don't really have ways to get that data into the public health system so we can calculate a positive predictive value, we have those data in our labs so we can calculate that and use PPV, positive predictive value as a measurement of newborn screening programs effectiveness. When we first started developing the quality indicators, this was the quality indicator that people were thinking, "Ooh, this is the one we really need to use." Positive predictive value is the most important, and while it does have some value, it's not necessarily the most important.

While we are having those conversations here some real quotes that I got from maybe some of you who are on the phone, and it's too hard to find false negatives, where are they, we know about the missed cases, but I've already closed out my reporting for that years. I follow kids in my clinic who were missed on the newborn screen, but I just haven't had time to call the state to report them. Positive predictive value, it's the best we could do, we can't report anything else, so it's just been some challenges with missed cases and I think there's also the issue of, they say we've missed cases, does that put our dirty laundry out there?

I would like to, so in the next couple of slides, reminding us where we get the false negative rate and how it can be manipulated through different ways that we develop our screening tests. So false negatives are those who have the disease who test negative on the screening tests, and if you'll all remember from your basic epidemiology classes, this is a screening test and we have A, B, C, D and E cells who are the gold standard for the disorder, those who truly have this disease on the top, the positives there, so the A's are those who have the disorder and test positive for the disorder on the screening tests.

The D's are those who do not have the disorder and they truly test negative. The B's then and C's are the false positives and false negatives.

The false negatives are those who have the disease who test negative on the screening tests so that's C, those who test negative, but truly have the disease over A + C, this also could be thought of one minus the sensitivity. So positive predictive value then is a proportion of true positives among all positives tests so that's the true positives, that's the A's over all of the positive tests, which is at A+B. So both of those depend on A, those with the disease who identified by the screening tests.

So getting back to our fishing scenario, we want to cast the net broadly, but no too broadly. So if you can see the darker colored fish there, those are the fish who have the disorder that we are trying to identify in newborn screening and the lighter color fish are the normal kids who do not have the disorder. In the situation that I depict here we have cast the net so broadly we have captured all the kids, they've all screened positive and we've captured all that have the disease, but we've also captured a lot of them who are normal and do not have the disease.

So, in identifying babies with the disease, this is about the same scenario and I think some animation has been lost in the translation here so I'll just talk through that. We have all the babies with the disease, so we cast that net so broadly that we have zero false negatives. Everybody tested positive for the disease so there were no negatives at all so no false negative. So our false negative rate, C / A + C is zero, zero percent.

The positive predictive value, however, is A / A + B and that is 0.7%. So that is very low and probably not an acceptable false positive predictive value for our test. We want to figure out how can we manipulate that a little bit. If we were able to take all of the babies and group them in some way that our asset was able to put them in the corner that we could say, "Yes, all of these babies are very similar." Then we cast that net, we could cast that net just exactly around those babies and have a very specific test to identify those babies.

So in this case, every baby is caught. All the fish are captured in that net so the false negative C / A + C, those babies were missed, 0 / 7 so zero percent, we capture them all and the positive predictive value is 100%, we were able to really discriminate the two cases for the babies, which this is the ideal situation, this is what we all strive for. However, very few tests actually work out this way. So a more realistic screen is where we are able to discriminate most of the babies, we can most of them, identify most of the true negatives as being true negatives.

You don't need to be come in for the testing. We can bring in most of the kids who really have the disease and say "Yep, you truly have the disease. We need to come in for further testing and follow up", but there's a couple other of those lighter colored fish that slip in there, fish that we need to bring them back in and have further testing and say "Oh, you know, your baby does not have the disorder that we're screening for". So there's very few of those babies that get in there and yet they do impact our numbers and there's one baby out there, that are dark colored fish that we missed.

So our screening test on this case, we identified six of the seven babies who truly have the disease, correctly, and the other one there was a false negative, that gives us a 14.3% false negative rate and then our positive predictive value was 10.7%. So, here we have a, and I've given a talk to some of you about the challenges of positive predictive value as well in other situations that we can talk about a little later, but that is [inaudible 00:20:30] newborn screening program and the boss of that program says I really want to maximize positive predictive value. That's the most important thing we want to do, so I said, alright, we'll maximize positive predictive value, we can do that, and here they've maximized it.

Every baby that tests positive on the screen, every baby they capture inside that match truly has the disease. So, we captured all of those babies and we have a positive predictive value of 100%. However, we've missed many, many of the babies, but if we just focus on positive predictive value and not the false negatives, we're going to miss those babies. So that false negative rate here is 57.1%, which we would all, I think, agree that that's far outside our acceptable range for what we'd like to miss. So if we go back to this more realistic screen and this really could be a cystic fibrosis strain in states, depending on your algorithm, we might have a 10% to 15% false negative rate. What you want to know then is why was this child missed.

So if you have the numbers and you know that baby truly was missed and you've identified him you could go back and say what was it, is it our assay, is it something with our follow up? Was the baby missed because our assay called it up, identified the baby correctly, but we didn't do appropriate follow up. What happened? You can do some quality improvement of quality assurance there. Some missed cases really do happen. The problem is we don't know the rate with which they do happen so why are there missed cases?

Well there is true biologic false negatives, those whose value is truly below the cutoff of the test. There are sample mix ups. They are rare, but they do happen where the sample mixed up, something got plated wrong, something happened and the baby got missed on the screen so maybe their value was called out as positive, but it was assigned to another child. There are laboratory errors. Something happens in the laboratory with the assay and proper procedures were not followed.

Something happened with that and there was a laboratory error and that baby was missed, or perhaps no newborn screen was collected. That's not a false negative, that's not a fault at the lab, the parents may have refused, something may have happened, it was a home birth, didn't get collected, whatever the situation may be, no newborn screen as collected so those cases are missed. What do we do about that? Where can the missed cases be found?

They can be found in the sub-specialty clinics in our local hospitals, primary care offices, for some of the disorders in some of our states, birth defects registries, more common now for CCHD but even some of the other disorders can be identified that way depending on how your birth defects registry is working, through hospital records, through disease registries, through death certificates. So, there's many places where those data exists. However, that's not an easy place to get the data.

It's not a trivial situation. So, let me give you a couple of biologic examples; child with cystic fibrosis, a baby with an IRT less than the cutoff that presents at six months of age, after struggling with weight gain, has some respiratory problems. Will this baby ever be seen? The baby was likely to be seen at the CF center bit will they ever let the state know that yes, here's a baby that we've identified? Some places will, some places won't. Is there a system in place?

So, MCAD, you can have a well fed or anabolic infant with MCAD and that baby may then have normal newborn screening results. If they did or they missed their newborn screening and it's left untreated, eventually that baby may get sick and die undiagnosed. Will the newborn screening ever know about this baby? So, how do we solve this problem? First thing is just, have an awareness of the incidents of cases, of the incidents close to what you anticipate.

So, are you seeing the number of babies that you anticipated that you should be seeing in your population? For many of us, that's, the numbers are very small. So, it's very hard to calculate this. It might take calculating it over a several year period to see if it's something close to what we'd expect. We need a communication system with some specialty clinics. What most of us have is communication with a sub-specialty clinic, where we can call someone and they will tell us; yes, we've identified a case you didn't know about back in forth. It's our only system, it relies on a relationship between people.

Then we need to develop relationships with primary care physicians and out of state and regional programs, partner with birth defects programs and then, finally, we can search death certificates for many of these and see if there are disorders that might be related to a newborn screen disorder. So, we need a surveillance program with protocols to identify and report the false negatives. This is not something that's trivial and many of you heard me say that by the time I retire I want us to have solved this problem, that we, really, will have a system that will help us identify these babies and then find out why were the babies missed.

We have some common definitions that we have built into our NewSTEPs data repository; were they biologic false negatives; where there errors; or were they not screened. By improving the newborn screen; we can really only improve this newborn screening if we really look at all of the outcomes. Positive predictive value is one teeth of the newborn screening that can be very helpful for us, really, at the laboratory level but we really need to have a good grasp on the missed cases. So, that puts the context of why we're looking at this false negative issue and I think I'm handing it back to John to introduce our next speaker who will be talking about a solution to this problem.

So, I've posed the problem and Arizona has a nice solution for that.

Thalia Wood: Hi, this is Thalia. I don't know if John did the star 7 to unmute.

Brigitte Dufour: Can you hear me?

Marci Sontag: Yes, Brigitte, we can hear you. So Brigitte is from Arizona and we will let her introduce the topic and she has a very nice solution into what they're doing in cystic fibrosis.

Brigitte Dufour: So, good afternoon, everyone. My name is Brigitte Dufour. I'm case manager for the office of newborn screening for the Arizona Department of Health Services and before I go further, for those hearing a French accent, do not adjust the audio of your computer or of your phone. I do have an accent. I will not let you guess of what country I am from. I will let you know right away; I'm from Quebec in Canada and I moved to Arizona in '98 and I still have that thick French accent. So, bear with me during the presentation.

If you have any questions I'll be happy to give you more information after the call or just raise your hand. So, the presentation of today is the objective of trying to understand the value of using a secular portal for communication with sub-specialist to recognize the benefits of using Sure Point for data sharing for presumptive positive confirmed cases and probable false negatives with sub-specialist and identify the benefits and limitations of using Sure Point.

Okay, so, just to give you a picture of our contracted sub-specialists in Arizona; for cystic fibrosis we are working with two CF centers. One is in Phoenix at Phoenix Children's Hospital and the other CF center is in Tucson with the University of Arizona Medical Center. For metabolic genetics, we are working with Phoenix Children's Hospital. For endocrinology, we are working with three centers; we are working with Phoenix Children's Hospital, with South West Pediatric Endocrinology and with the University of Physicians in Tucson.

For etymology we are working with the Center for Cancer and Blood Disorders at Phoenix Children's Hospital and also the University of Physicians in Tucson. So, let's get back with the CLSI guidelines in regards to newborn screening and false negative. So, the new guidelines released in May 2013 states: "false screen negatives and false screen positive findings occur for various reasons including biological variation, limitations of testing methodologies and procedures, communication or other issues with the newborn screening system.

To evaluate the risk of a false screen negative finding and the reasons for it, screening programs should actively seek to identify case diagnosis outside the screening system using public health reporting system and sub-specialists or primary care providers" and this is the subject of my talk today; how we, in Arizona, are using public health reporting system to capture all this information. So, false negative in newborn screening,

and Marci did a really good presentation about that, it's a challenge to accurately calculate the missed case rate without a system to collect the data.

On our end we're using a portal called Share Point and it's a useful tool for the newborn screening program to collect and share the data without sub-specialists and also Share Point is valuable for communication and reporting of possible missed cases and we encourage discussion among our specialists to improve their newborn screening system. So, this slide is a screenshot of our portal.

This portal was created in 2002 and that portal was created following the September 11 event. It was basically a public, sorry, it was an emergency preparedness portal. We had, at that time, so many calls for suspicious powders due to anthrax scare. We had following, later on we had the West Nile virus outbreak. Then we had the H1N1 and all that preparedness for public health. So, that portal was put together to have all the country health departments together, law enforcement, other officials. So, we all have considered the response to those events.

So, what that portal is, it's public health professionals use the portal for, to search information and documents, to create alerts to inform the other user of what is going on and the next two points; number three and four, this is why newborn screening is now using that portal, is because we can manage documents, store in the document library, we can upload and edit spread sheets containing the patient information. We can track and review version of other documents such as diagnosis form and we can share patient care information in a secure environment.

On top of that with that portal, we have technical support Monday to Friday from 08:00 am to 16:30 by phone or by email. So, it's very easy for the user to just log on into that portal. The sub-contractors that I was talking about, they all have, not all of them but the cystic fibrosis people and the metabolic genetics team have access to the portal and they need to sign a confidentiality agreement to be able to log in into that portal.

So, this is Share Point on the top of the slide. What we did and widened out circle in 2010, we decided to piggy back that portal and we created a sub-site for newborn screening. So, what we did is on the lower right corner of your slide, we created several sub-sites for cystic fibrosis, endocrinology and metabolic disorders. So, this is a screenshot of o9ur

website for cystic fibrosis. So, both CF centers in Phoenix and in Tucson have access to the portal.

What we capture is all our presumptive positives, all those patients with their RFC levels, their sweat test results and we also capture, unfortunately, two cases that were missed but we had that thread of discussion and we were able to tweak out algorithm or to correct things that were going on in our system and to better screen those babies. With the metabolic group it's a little bit different. What we are using with that circle portal, we're using the secure email notification. So, all our communications including calls from pediatrician or inquiries from the ornithologist are recorded in those email thread and what is beneficial of that is we can just cover copy all the follow up team because we have three other follow up specialists here.

So, whomever is getting the call will go into Share Point and notify everyone about the call we got from a concerned pediatrician about a baby with some symptoms. We have some challenges in Arizona. For example, we have a huge Hispanic population, American Indian population with different genes. So, that may also be a reason why it's always important for us to keep in touch with all our specialists about what's going on and we have, also, unfortunately, babies that move out of state or to Mexico that, unfortunately, we don't always know the outcome of those babies but this is all captured in our Share Point.

So, the benefit of using that portal is it's a very robust portal, established in 2002. We have strong IT support. We are able to share presumptive positives and confirmed cases and possible missed case with contracted specialists from different clinics and centers and it's a shared knowledge between specialists. Everybody can learn from others what is going on in the state.

The limitation of using that portal; you need IT support because, unfortunately, sometimes, a user will lose the password or they're unable to log in. So, they need to call the help desk. You need some training with your specialists, you need to send them the link, you need to tell them where to go and where to put the information and why it's important for them to log at least once a day into that portal to find more information.

You need to keep the site neat and tidy so it's easy for everyone to find the information they're looking for. You need to eliminate, sometimes, a duplicate spread sheet so nobody is using the wrong spread sheet when they update a patient and the success is depending of data sharing. We all need to get feedback from the specialists about what they found, what calls they got from physician or concerned parents about their babies. If there's no communication both ways, well, then there's no need to put a Share Point site.

That was the purpose of my talk today; to talk about using a portal to capture all the data and also to encourage communication with your sub-specialist. My name is Brigitte with the Arizona Department of Health Services.

Thalia Wood: Thank you so much Brigitte. Marci, did you want to go on to the last topic or do you want to open up the call to questions at this point?

Marci Sontag: I would say John and Carol that's chairing this group, I would lean toward, if John and Carol agree, I would lean towards, let's get the questions out about false negatives and then we can spend the last 10 minutes or so talking about the last portion of the call.

Thalia Wood: Okay, so, why don't you unmute everybody's phone lines?

Marci Sontag: Okay.

Telephone: This conference has been unmuted.

Carol Johnston: Hi everybody, this is Carol Johnston and I wanted to take a moment to very much encourage anybody who has any kind of false negative policies to send that information to Thalia to be distributed to our entire group. Her email is Thalia; t-h-a-l-i-a dot Wood at APHL dot org. Alright, and I think, with that we'll [inaudible 0.38.13].

Thalia Wood: Well, there were a couple of questions that were written into the chat. One was for Cindy in Vermont. Tell me where are your specialists set for screening, Cindy?

Cindy Ingham: Can you hear me?

Thalia Wood: Yes.

Cindy Ingham: We use New England Newborn Screening Program lab, Austin, as does Maine, New Hampshire and Rhode Island.

Thalia Wood: There's a lot of background noise, I don't know if somebody is out there that can mute your phone but there's a lot of background noise.

Cindy Ingham: Thalia, is it just star 7 they press on mute?

Thalia Wood: If you want to mute everybody and then they can push star 7 to ask their questions. That would be fine.

Cindy Ingham: Okay, because there really is a lot of noise. So, I'm going to go ahead and mute everybody. That's star 7 when you'd like to talk.

Telephone: The conference has been muted.

Cindy Ingham: The other question that was asked on the chat box is to Arizona; can all specialists see all of the cases posted on the Share Point site?

Brigitte Dufour: Hi, this is Brigitte, can you hear me?

Cindy Ingham: Yes we can.

Brigitte Dufour: And if I click on that ... Let me get back to the slide, bear with me. Okay, so if you see those sub-sites for cystic fibrosis, endocrinology and metabolic disorders, so the cystic fibrosis specialists will only see the cystic fibrosis patients. They won't see anything about endocrine disorders and the same thing. So, they will only see the patients that pertain to their specialty. Does that answer your question?

Cindy Ingham: I think that does. I'm speaking to Louis and she might not be on the phone, she's just on the web but I think that there's always that concern of privacy and people think, that hit that issue, of seeing patients you're not supposed to have access to.

Penny: So, this is Penny [inaudible 0.40.19]. So, if you have more than one center for a given condition can all of the centers see kids that are residing in let's say, the other centers' geographic region?

Brigitte Dufour: Yes.

Penny: Okay, so all specialists of a given specialty can see all cases?

Brigitte Dufour: Correct and keep in mind with the contract they have, they already have a confidentiality agreement and a pledge to protect the information and on top of that by using that portal, they need to sign a third document which is the user agreement for Share Point. So, we're really trying to protect this information and they will only see patients with their disorder.

The benefit for that, I will give you an example, for those babies born in Southern Arizona, like in Yuma, we never know if they will show up in Phoenix or if they will show up in Tucson. So, by sharing those presumptions results to both centers, whomever will get it will put that on their spread sheet and we know the results. So, it's better communication and we know where the babies are at. I will unmute now.

Thalia Wood: Thanks. Marci, do you want to go ahead with the other questions in the chat box?

Marci Sontag: Sure. Wow! There are other ones coming in. So, Kimberly Piper's asking; is anyone working with Fetal Infant Child Mortality Review programs to find potential missed babies. I'm hoping someone's just unmuting their phone and can share their experience with us. I think we'd all like to know.

Cindy Ingham: This is Cindy, can you hear me?

Marci Sontag: Yes we can.

Cindy Ingham: Fetal death review committees sends me copies of all infant deaths up to the age of about 15 months and I also get copies of death certificates on all babies of that age range. So, I can look through, pull the newborn screening, talk with the medical examiner if necessary. So, it's more of a notification. I'm not on the review team.

Marci Sontag: How many cases would you say, in your state, that come back to you if you're seeing all infant death certificates?

Cindy Ingham: Under 20.

Crystal: This is Crystal in Nebraska and we give a list of all the confirmed positive cases that are identified in any given calendar year to the child death review team coordinator here that also is in DHHS.

Male: Hi, [inaudible 0.43.16] can you hear me?

Marci Sontag: Yes, we can hear you.

Male: We have a child death review, they [inaudible 0.43.23] probably because they, and I think in Marci's talk the last point [inaudible 0.43.31]. So, we get feedback from them but it's not predictive.

Marci Sontag: It definitely seems like that's an area that, with the right system, could be helpful but I think there's a lot of work that needs to be done to help make it helpful for all of us. Anyone else have any experience with that that they'd like to share?

Harry Hannon: Is the line open for questions yet?

Marci Sontag: The line is open for question.

Thalia Wood: It is.

Harry Hannon: This is Harry Hannon and I have an issue with terminology that's been used; we're calling everything missed cases instead of delayed diagnosis. We don't really know if they were missed or not until we do a gap analysis. So, actually, they are delayed diagnosis. If you truly missed them, you probably wouldn't know they ever existed but a missed case is implied there was an error in the system and then one has to do a gap analysis to identify if there was an error.

It could have been a metabolic justification and you couldn't have detected it at all. So, if you call it a delayed diagnosis and then investigate, then you can declare that it was missed because of a particular reason but calling them all missed cases implies lots of errors in the system which may not be true. They're actually detected by a diagnosis outside of screening and they're presumed to have been screened and then you investigate that they were screened and now you do a gap analysis to determine what happened in the system that they were not detected.

So, I know this terminology gets a little confused when we call them missed cases but if you look at most of the things that we'll say that they are delayed cases "missed" and not the other way around. So, I'd appreciate anybody else's comments on this use of the terminology. I know we have a [inaudible 0.45.37] project to deal with standardizing

terminology but it's just underway and that and screening positive and all assorted issues, there's a lot of terminology that we float around in different ways for different meanings.

Thalia Wood: Thanks Harry, this is Thalia. I think that's a really good terminology you used; delayed diagnosis. What do others think about that?

Marci Sontag: Yes, Harry, that's an excellent point and that's how we refer to it in our repositories as well is really asking the question; was this baby diagnosed after the newborn period and then what was the cause of that diagnosis, what brought them to attention. So, I think that's an excellent point and we all need to be more careful with that. Any other comments?

Thalia Wood: Any other comments? Yes, don't forget to unmute your phone, star 7 to ask a question.

John: Brigitte, I have a question. It's John in Washington. I understood that your sub-specialists like for example cystic fibrosis would be able to see all of the positives that you have reported out and referred for diagnostic testing and then at some point they're going to be associated with one or the other of those two clinics. Will your database reflect that they've been assigned to one of those clinics or they've chosen to be seen at that clinic?

Brigitte Dufour: Yes. So, usually they will be assigned to, depending where moms reside, either to the Phoenix spread sheet or the Tucson spread sheet but they both can see either way, spread sheet. Yes, they are assigned to centers and they are able, from there, when the baby is tested, to put the sweat test results and give us also a confirmation for CRMS or ... Yes.

Marci Sontag: Does that answer your question?

Debbie: Brigitte, this is Debbie Freedenberg, are you saying they're using that Share Point to obtain the confirmatory testing results?

Brigitte Dufour: No, I'm not. No, I wish. No, it's a way to gather the information and to keep the history about those babies and all those data collections, IRT value, birth weight and all the information and to share with both CF centers but I still do that phone calls, faxes, contact the pediatrician to find out if the baby has been referred to the CF center, which CF centers and things like that. John, do you have any other questions?

John: No.

Thalia Wood: Okay, well, we have about 10 minutes left. Marci, do you want to go ahead and give your little talk here?

Marci Sontag: I will. So, the title of this slide here is the APHL response to the Milwaukee Sentinel article and I'm actually going to hand it over to my colleague, Jelili Ojodu to first talk about the APHL response and give some background on the broader responses then we'll talk about it a little bit from the NewSTEP's perspective as well. Jelili are you on?

Jelili: Can you hear me?

Marci Sontag: We can hear you.

Jelili: Alright. Good afternoon, everyone. So, this topic is, I'm sure, very familiar to the folks on the phone. Over the past several months you have gotten our requests by some of the reporters for the Milwaukee Journal Sentinel to collect information on a number of things. What they ended up focusing on at the end of the day was the transportation of specimens from the hospital to the laboratory and how that differs from state to state or hospital to hospital.

An article or five part article was written and published on November 16th and it highlighted among other things the differences especially highlighting hospitals that do not tend or hospitals where samples are not received in a laboratory after five days among other things there. So, we at APHL have done a number of things to try and work with our member states' public health programs in general to address this, I think, major deficiency that was brought to our attention as it relates to this particular article.

One of the things that we did was right a number of talking points, message pallets that we distributed to newborn screening programs and state newborn screening laboratory directors. We've held a number of strategic calls with ASTHO, AMTEC, Association of Maternal and Child Health programs, the March of Dimes as well as the American Hospital Association to figure out how we can work together to address these kinds of issues, whether it's, the article did note a number of things and suggested, among other things, states should be open seven days a week, every state should have a courier system and all of those kinds of things.

If these were to be instituted there will be a number of things that would have to go in place depending on the state that we're talking about here. We have had a number of media coverage analysis and also media exposure related to this. In fact, the reported has interviewed our executive director to talk about our response and how we're working with our states to address these and most of those have been posted on a number of places including the NewSTEPs website. So, feel free to check that out on our website and the response from the association on there as well.

We have sent out a survey working with the Discretionary Advisory Committee of the Secretary as relates to newborn screening and heritable disorders on transit time for the specimens that are collected from the hospital to the laboratory and we've gotten a number of responses back from states. We have not gotten some responses yet from other states who have requested an extension. So, if you're on of those states that haven't responded yet, please, respond to that survey as soon as possible. If you need an extension let us know and if you don't know anything about this survey in question, feel free to call me or email me after this call and we'll definitely follow up with you on that.

Then, there are a number of things that we're planning to do in collaboration with the number of sub-committees and committees that we're working on including the development of a policy statement on this particular issue, working with AHA, the Hospital Administration to develop a webinar, a joint webinar that would encompass the path of the spot from collection to the laboratory and then reporting out those results and then, among other things, developing the number of educational activities that we can work with our members on and to address or highlight some of those activities especially as it relates to NewSTEPs.

I'm going to pass it back to Marci to talk about what continuous quality improvement, talk about the psych assessment and evaluation that we do for different states that we plan to do in the coming months for a number of states and all of this fits nicely into what a comprehensive newborn screening resource center provides to the newborn screening community as a whole and so, Marci.

Marci Sontag: Thanks, Jelili. The response to this from the NewSTEPs perspective; this is really our bread and butter and, really, what we are formed to do. We

are here to support state newborn screening programs and help with quality improvement. So, as Jelili said, there are many components to this. The first, that I think all of you have heard about through our data repositories, we have a list of quality indicators that have been developed by state newborn screening programs with input at many different levels by the state newborn screening programs and other stakeholders in newborn screening and one of those, for example, one of those quality indicators is directly related to transfer time.

Transfer time, testing time and time to get the results back to the PCP's and to the specialists. So, these are the exact types of things that quality indicators exist. We have been now funded for slightly over a year and a half, the data repository's ready to go and so, these questions, when they come up in the future, we will have those data and our goal is to say; "ooh, you know what, we notice that state X has some outcomes that are slightly different than the rest of the country". The state will have their profile and we'll be able to see how they compare to the rest of the country but then we are here to help that state identify the resources to move those outcomes to be closer to where the rest of the country's outcomes are.

So, that's really what we are here to do. We've been spending, really, the past year and a half developing the data repository and the outcome measures. Now, we are ready to accept data and to have some understanding, we can accept that data and these problems will not be surprises to us. We will be able to say: "hey, we have this technical assistance program that is here to really help and help move forward". Other aspects of that beyond the data itself and then our response to how to give you those reports.

So, as we said, we have site visits that we have a comprehensive site visit. Many of you know, New Jersey was site visited last spring. We have a couple of site visits with other states coming up in the coming months and those site visits, really, are ... We bring in experts from around the country that help identify what the challenges, what are the strengths of this state so we can help you problem solve some of those challenges and use the strengths in your given state to help other states as well.

We have other things in the works that we're working with some of the regional collaboratives to help develop mini sites as a tool that you can do in your own regions to help with those types of things. So, this article

has just highlighted the importance of what we're doing and we're really excited to be able to move forward so these types of things, when they come up in the future, will not be surprises. We'll be on top of it and be able to support you through any of the challenges that come forward.

Jelili, do you have anything else you'd like to add?

Jelili: No, nicely stated. If there are any questions on the phone it would be nice. We certainly are here to listen and if you want to email us any questions you have or comments feel free to do so.

Thalia Wood: Absolutely. Thank you, Jelili. If you have any questions, don't forget to do star 7 to unmute your phone. If there are no questions, Carol, would you like to wrap this up since we're getting right to the top of the hour? Carol, did you unmute your phone?

Marci Sontag: I will step in for Carol. I'd like to thank Brigitte so much for your presentation on your approach in Arizona and Cindy for presenting your state profile for Iowa and please, if any of you have any approaches to false negatives that you'd like to share with others, programs, we really include you as Thalia said earlier, please send those ideas to Thalia as well as if you have other suggestions for topics for future webinars, we would love to hear from you.

Thalia Wood: Absolutely. I will be sending out a brief survey and we definitely appreciate your responses and actually the state profile was in Vermont, not Iowa, so, yes, thank you Cindy for that.

Marci Sontag: Did I say Iowa?

Thalia Wood: You did.

Marci Sontag: I'm so sorry (laughs).

Thalia Wood: Anyway, thanks again, everybody. So, we'll be doing another call in a couple of months and we're sending our more information. So, Marci you can stop recording then, thank you.

Telephone: Thank you, please stand by.