NewSTEPs

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Short Term Follow-Up Technical Assistance Webinar

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Presentations: Sharon Linard, MS, State Ohio NBS Profile

Felicia Wilson, MD, Hematologist in Alabama

Lianne Hasegawa-Evans, CGC, Kapiolani Medical Center and formerly with the State of Hawaii Genetics Program

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

Sikha: Sharon, John and the speakers, you may dial star seven to begin speaking.

John: Fantastic. Thanks Sikha for taking care of getting this rolling. I like to welcome everybody to the first Short Term Follow-Up Technical Assistance Webinar of 2016. Glad to have you here today with us. If you have colleagues that are unable to be with you today watching this, this will be our archived and will be sending out an email in the near future when that's available with the hyperlink for you. We'll start today's topics about Hemoglobinopathy follow-up. Looking forward to hearing the presentations. I will introduce the speakers.

Before we get to the main topic, we will have states profile highlight from Sharon Linard from the State of Ohio. We do this at the beginning of each webinar. It's not related to the topics but it's an opportunity for this individual states to show some of the things that are happening in their programs and teach us a little bit what they do and why they're unique so that we can learn from each other. It's been very helpful in the past. We're looking forward to hearing from Sharon.

Following her presentation, Felicia Wilson from Alabama will speak to us. She is positioned there and involved with the hemoglobin disorders, and taking care of babies with them. We will have our final speaker will be Lianne Hasegawa-Evans is the genetic counselor formerly with the Hawaii Department of Health Newborn Screening Program and that she have a lot of good experience

understanding their program and their use of telemedicine to help with the Hemoglobinopathy follow-up. We'll turn the time to Sharon.

Sharon Linard: Can you hear me?

Sikha: We can hear you.

Sharon Linard: Okay, thank you. Hey, I'm here with the follow-up staff. There are four of us here in Ohio who do a follow-up for newborn screening. You can go ahead and go to my first screen. We have three very distinctly different programs in Ohio, the congenital hearing loss or universal hearing screening, the congenital heart defect screening and then the blood test screening that are all separately managed and do not share data. I am here primarily to talk about the blood screening. This is Ohio's 50th year of screening. Our anniversary is July 1st just when in 1966 is when screening began in Ohio. Next slide.

Just in summary for our hearing screen, we've been doing screening in Ohio since 2013 or since 2004, and this is the summary data the program shared with me for 2013. They're always about a year in [inaudible 00:03:26] their data management because of how they collected on the birth certificates, so you have to wait until the birth year is finalized. Next slide.

Our congenital heart defect testing is still way … began in 2014 but we are still enrolling hospitals and getting them all on board. By the end of 2015 we basically have all the hospitals ready to go and are really going to start our first year of complete testing with 2016 [tailing the 00:04:07] 2015. In 2015, we did diagnosed two babies with severe congenital heart defects. Next slide.

In Ohio, we test for the 36 disorders that are part of the recommended national panel. Next slide.

We in an average month have about 11,600 screens that we received, 95% of those are collected in the first 24 to 48 hours of life. We result out 90% of our samples within four days of receiving them at the lab, and 88% of our initial samples are reported out by the seventh day of life, and then average month we have about200 repeat screens. We do not encourage repeat screening in Ohio. We have about 20 unsatisfactory samples due to quality and about five religious objections. Next slide.

In a year we have about 140,000 samples, about 3,000 of those resulting in abnormal call for disease risk, another 3,000 have issues of inconclusive results because of transfusion or early draw drawn before 24 hours or misinformation, and then we have about a total of 200 unsatisfactory samples. Next slide.

This is a summary of our data since we began based on disorders. In the last 50 years, we've identified over 4,700 babies with diseases that have benefited from early diagnosis and treatment. Next slide.

In 2016 by legislative order, we are going to begin Krabbe disease testing, assuming we can get the equipment and changes to our database in place. Right now, the date the legislator gave us was … that we were to begin screening on July 1st. However, they just approved our capital budget to purchase the equipment today, so it's going to be a real price raised to see if we can actually get the equipment and validation done and everything by July 1st. We also are hoping at the … We are using the mass spec to do Krabbe testing and we are hoping to do Pompe and MPS1 validation at the same time and go live shortly after we go live with Krabbe. -XALD is probably a little further down the road but also one that we plan to add fairly soon. Next slide.

The thing that we are working on here apart from all states are looking at timeliness but we have discovered that there is one primary area that we need to work on. If you look at our data from 2015, it's pretty consistent as to how long it takes samples to get to the lab. We pretty much get 57% of samples in the first 48 hours after collection, but we have this 14% that are hanging out there and are taking a long time to get to the lab. It's not real obvious looking at the data based on this graph as to why those samples are late. Next slide.

If you actually break the data down based on the days of sample was collected, so along the left side is the date the sample was collected. It becomes pretty obvious that the orange and red samples become a problem for samples and that samples that get here after 48 hours. That's the problem for samples drawn on Friday, Saturday, and even Sunday to a certain extent. What we are looking at doing here at the lab is we currently have a courier that it runs six days a week. They'd pick up samples and drop them at the lab Monday through Saturday. We're doing a variety of different things to try to address this for weekend shipping, one if we are visiting all of our hospitals and talking with them about what barriers they are facing that keep them from shipping over the weekend.

We also are changing our shipping so that samples are picked up at the hospitals Sunday through Friday and delivered to us Monday through Saturday, so that the courier is not holding on to samples for two days on the weekend because of not running on Sundays. We're hoping that also will help with this timeliness issue. Other than that, a lot of it is just changing culture at the hospital so we're hoping to do that through our site visits and through where we've developed a report that we'll go out quarterly to the hospitals so that they are kept up to date on exactly where they stand on this. Next slide.

I want to thank you all and I want to thank everyone working in newborn screening. This is a quote from our parents, one of our parents, the first baby we diagnosed was in Ohio. I think her thanks go out to everyone working in newborn screening. That's all I have.

Sikha: Sharon, this is Sikh. Thank you so much for that overview and especially about last night, it really is heartening to see this feedback and it really does, it make

us feel great about what we're doing and certainly what [inaudible 00:11:08] are doing. Congratulations to you on that. Nice job.

We had a few questions that we can address right now really quickly before we move on. The first one was, you noted one of your slides that you have 20 unsatisfactory screens rejected due to quality, and the denominator for that month you noted was 11,600 satisfactory initial screens. The question was, are those 20 unsat for the month or are they total for the year?

Sharon Linard: Therefore the month, we have about 20 per month.

Sikha: Okay, great. The other question that we had in the chat box was regarding Krabbe screening in Ohio. Do you use the tandem mass spec platform or the advanced liquid logic platform in that screening?

Sharon Linard: We will be using the tandem mass spec and that is interesting how the whole thing arose, but the Senator actually placed the wording in the state budget and state budgets cannot be … they are up for discussion, they aren't up for debate. The wordings specifically said that we must use mass spec and we cannot do second tier DNA testing.

Sikha: Thanks for that feedback. We're all shaking our heads in the room here and we'd love to hear a little bit more about that and maybe [inaudible 00:12:38] talk to you to get more information on those limitations within your legislation. With that, so I will turn it back over to you.

John, a bit honky. We can't hear you.

John: Yeah. We're ready to go with Dr. Wilson. Thanks.

Felicia Wilson: Okay. Can you hear me now? Hello?

Sikha: We can hear you. Yeah.

Felicia Wilson: Okay, great. My screen just blacked out. Okay, let me pull it back up. Here we go. Okay, I'm going to talk about newborn screening for Hemoglobinopathy in Alabama. The next slide has my financial disclosures. The next slide, newborn screening for Hemoglobinapothy was initiated in Alabama in 1988.

The next slide, we show the statistics in this bar graph for five years denoted at the bottom of the graph from 2009 up to '15. We diagnosed between 33 to 62 babies each year. The 33 is an outlier so usually when we look back over the day those were the 27 years that we've been screening. We do see somewhere between 50 and 60 babies identified with sickle cell disease.

The next slide shows the network that we have in the state which has been very successful for us. When a baby with a positive screen is identified, of course, the

parents and the primary care provider receive a letter of notification, but we have two major comprehensive sickle cell standards in the state that provide care for these infants. According to geographic lines, the letter of notification also goes to the appropriate center which is either University of Alabama in Birmingham or University of South Alabama in Mobile. The color-coded map denotes the seven regions that the state is divided into and each region has the community-based organization that also helps to enhance services and compliance. These community-based organizations provide social support to the client, and also have the memorandum of understanding with Alabama Department of Public Health to provide counseling for those families who have babies identified with trait and not the disease.

All of these entities are then governed by a state oversight regulatory commission to make sure that we are meeting our goals. For instance, when one of the centers is notified of a positive screen, the nurse coordinator is responsible for establishing an appointment for that infant within 72 hours. Then, of course, the commission oversee that our success rates are really high in getting these babies in because the goal is to start prophylactic penicillin by three months of age. Our success rates have been greater than 95% but I think we have a very coordinated effort here in the state to get the babies in.

The next slide talks about some of the challenges that we have in screening. One of the challenges in screening is getting specimens from babies with extreme prematurity. In this case, the only hemoglobin they are making yet is fetal hemoglobin, so we are not able yet to see the sickle hemoglobin that would then aid and diagnose this for these babies. We do need follow-up for them but, of course, a lot of our babies with extreme prematurity may require blood transfusion during their stay in the NICU. Blood transfusion prior to screening is problematic because then the test that we're using which is high-performance liquid chromatography is actually done on donor blood and not on baby's blood. For these scenarios we do the screening upfront but we ensure that we get a second more accurate screen in about three months, two to three months when the donor blood hits [inaudible 00:17:13].

The next screen addresses challenges we have in notification. The biggest challenge is having incorrect demographic information so that by the time the nurse coordinator tries to contact the parent maybe the phone is no longer in service or we have an incorrect address which leads to delay in follow-up and initiation of care. A number of these babies will at some point later show up on the doorstep of a primary care provider. In that situation, sometimes a lack of understanding and interpretation of newborn screening also result in a delay of care. We have had a few cases where maybe the child was now three years or five years of age. We even have a 15-year-old where parents have been told the newborn screening was fine but it was a mild form of sickle cell disease that was not understood to be the case. We also have the parental acceptance and compliance. Particularly when the family had no knowledge that sickle cell trait

runs in the family then they may be hesitant to accept that this baby does have disease.

On the next slide, we look at challenges in data and statistics. While we know the number of babies that have been identified by newborn screening, the exact number of individuals living in Alabama with sickle cell disease is unknown. We know that some people move out of state, other people move into the state that may not have been tested in our system and, of course, mortality may account for some of that, so the impact of sickle cell disease on their health is not known. To overcome these challenges, we'll require newborn screening to expand relationships with all health care professionals and facilities to ensure the proper surveillance. This is actually been a problem nationwide which the CDC and the NIH have been working to develop a registry, but right now there are limited states that participate in that but that's one of the challenges that we have as well.

The next slide outlines the challenges we have in diagnosis. Although the primary purpose of screening is to identify infants with sickle cell disease, we also identify infants with other Hemoglobinopathies and Hemoglobinopathy care. Again, the lack of understanding and interpretation of newborn screening may result in some delay. For instance, those babies that have mild forms of disease like hemoglobin C disease or hemoglobin C beta plus thalassemia or some of the other thalassemias like E-beta plus thalassemia may be missed because it's not sickle cell disease, so there's a lack of understanding about the interpretation of that sample.

In addition, sometimes we get parents who've been told they have disease but what they have is [maybe 00:20:43] alpha thalassemia trait or a beta which is hard to diagnose during the newborn period. We don't see this problem too often but sometimes they are false negative and false positive. For instance, we may see a fetal hemoglobin only, so that could mean prematurity or it could be diagnostic of beta thalassemia major. In rare instances, sometimes in a baby who is term we do see fetal hemoglobin only. When the parents come in distressed then we do confirmatory testing, it turns out that they are normal. Those are the challenges and diagnosis.

One other thing I want to mention, so even though newborn screening is for detection of infants with disease so that early treatment can be initiated, sickle cell disease has been the center of very contentious and raging debates when it comes to challenges with sickle cell trait. The next slide kind of what I mentioned what those challenges are for sickle cell trait. It is important to notify these individuals of their trait status to enable them to make and form decisions about reproduction later in life and to also educate them about potential health outcome that might be associated with sickle cell trait.

In general, we've considered that sickle cell trait is the benign thing that doesn't have health consequences but more and more data suggest that there may be

some complication of sickle cell disease that can be associated with trait in extreme conditions like conditions of low oxygenation or extreme physical stress. While the seven community-based organizations are responsible for counseling parents of babies identified with trait, we often find that the information is not reliably passed on to the infant once they reach child-bearing age, so we still continue to get cases of babies identified where parent had no knowledge that sickle cell trait even ran in the family.

On the next slide, what we have are the statistics for sickle cell trait for the years 2008, '09, and '10, and these are pretty representative of the cases that we get over the 27 years. We see about anywhere from 1,700 to over 2,000 babies that are identified with trait. At the bottom of the slide, you see the racial makeup of those babies. While it is predominantly a disease of African-American, about 7% of samples that we test in the state which is about 60,000 a year, about 7% of those babies identified with trait are white and about 1% to 2% Hispanic as well as other races and ethnicities.

The next slide just mentions the diversity that we're seeing and with the growing racial and ethnic diversity of babies identified other than African-American, we need to be cognizant of that and make sure that we have medical literature written in appropriate languages.

The next slide is from a paper published by [SUSARs 00:24:31] and associates that looked at some of the definite associations probable, possible, or unlikely that you see in the medical literature associated with sickle cell disease. There's a lot of evidence to substantiate those definite association but, again, we have to approach those with cautions because such association do not necessarily prove [cause 00:24:57].

The raging debate is shown on the next slide with sickle cell trait in the athlete. The first case of an NCAA athlete of dying with sickle cell trait after extreme exhaustion came in 1973, and since then there have been at least 21 collegiate players where sickle cell trait has been linked to sudden death. The biggest cause of debate was a lawsuit that was filed by I guess the NCAA in the year 2006 after a player with sickle cell trait collapsed and died after sprinting 100 yards for the 16th time. As a result, the NCAA mandated that all division one college athletes be screened for trait unless they provide a prior test result or signed a waiver declining the test.

On the next slide looks at the NCAA policy since that time. Actually, a paper published showed that all of the sudden death related to sickle cell occurred in division one athlete playing football, but in 2012 the policy was extended to division two athletes and in 2013 to division three athlete.

In summary on the next slide, in 2015 in October, Alabama celebrate at fifth years of newborn screening. We had a conference where some of the individuals who have been diagnosed by newborn screening or parent

participated in that conference and it was an excellent conference that many of the parents shared the same sentiment that the parent in Ohio did.

Sickle cell disease is the most common disorder detected by newborn screening in Alabama. It is clear that while reports of association with sickle cell trait are alarming, the research studies a long term medical surveillance or paramount importance to answer the question to medical risks for the individual with trait and to institute evidence-based medical intervention. Despite these challenges, newborn screening for Hemoglobinopathy is one of Alabama's most important and effective public health program. Thank you.

That concludes the presentation for newborn screening for Hemoglobinopathy in Alabama.

Sikha: Thank you Dr. Wilson for that presentation. We had one question that popped up in the chat box and that is, what type of training do the community-based organizations received to perform the counseling?

Felicia Wilson: We do offer here a counselor certification or a counselor educations course here that is a joint effort by the Sickle Cell Disease Association of America, Mobile chapter and University of South Alabama. It's a nationally recognized conference. We generally have it twice a year and we get individuals from all over the United States to come for the counseling to learn about how they should respond to individuals with sickle cell trait. All of our community-based organizations are required to have people who have been certified through this course in order to be able to do the counseling.

Sikha: Thank you so much for that. Another question just came up asking, does Alabama supply NCAA athletes? Were there any one screening result?

Felicia Wilson: Yes, we do. In fact, we are trying to get a system in place to secure remote system in place where for Americare providers can access that information, but until now we do provide that. In addition, for those athletes in the State of Alabama who may not have been born here and don't have access to their newborn screening result, the Sickle Cell Association here in Mobile does provide screening free of charge for the general public.

Sikha: Thank you. John, can we go ahead and move on to the next presenter? Lianne from Hawaii? Yeah.

Lianne Hasegawa-Evans Hi, just so I'm here.

Sikha: Hi.

Lianne Hasegawa-Evans: Hi. I'll be speaking about our system of follow-up for alpha thalassemia and other Hemoglobinopathies in Hawaii. We faced several issues similar to

those that Dr. Wilson spoke about with sickle cell disease and trait in Alabama. Next slide, please.

In Hawaii, our State Newborn Screening Program began screening for Hemoglobinopathies in July 1997, and Hemoglobinopathies as a group in Hawaii are the most common screened positive conditions with alpha thalassemia being the most common. In fact, about 1 in every 42 newborns screens positive for alpha thalassemia. As you know, it's a common condition in Southeast Asians, so this makes sense given Hawaii's racial and ethnic makeup.

Ideally, families of newborns with a positive newborn screening result for alpha thalassemia are really any hemoglobin disorder should receive confirmatory testing, genetic counseling in management and appropriate medical management in education, and this is especially true for the more significant hemoglobin disorders like hemoglobin H disease. Next slide, please.

However, when we first started, there was really no protocol for following up on positive newborn screening results for alpha thalassemia and other Hemoglobinopathies, so pediatricians just try to use their best judgment. This often meant that pediatricians either provide the families with the counseling and confirmatory testing themselves. They refer families to specialist like hematologist or geneticist or really they just didn't follow-up the results at all. We recognize the need for more standardized follow-up procedures and we established the Hemoglobinopathy clinic in 2006 as part of our pediatric genetics clinic. Next slide, please.

That hemoglobin clinic, it was developed and is maintained as a collaborative public-private partnership with resources provided by both the Department of Health and Kapiolani Medical Center which is our children's hospital in the state. The newborn screening program provides funding to the clinic for in administrative assistant to handle the increased phone calls, referrals, and other people work generated by adding an additional clinic. It also funds alpha thalassemia DNA testing, and I'll talk about this more in my next slide. Genetic counseling services are provided by genetic counselors from the Department of Health Genetics Program. The medical geneticist from the pediatric genetics clinic provides supervision. She signs off on all cases. The Hemoglobinopathy clinic itself is housed within the pediatric genetics clinic which is part of the Kapiolani Medical Center. Next slide.

As part of our clinic, confirmatory testing is offered to all families. For other Hemoglobinopathies this would be like hemoglobin E or for us unknown hemoglobin variants. It usually includes just a simple CBC or at the most the hemoglobin electrophoresis, but alpha thalassemia really can't be confirmed on just the hemoglobin electrophoresis and DNA testing has to be done. Therefore, we routinely offer DNA testing for alpha thalassemia to both the newborn to help confirm their newborn screening result and really to determine the type of alpha thalassemia they have whether it's trait or hemoglobin H disease. We also

offer it to mom and dad and that helps us provide accurate recurrence information and genetic counseling.

However, about a year after our clinic was started, we started to realize that some insurance carriers wouldn't cover or would really only partially cover testing, and, of course, this affected the families' willingness to complete their testing and made it difficult for us to provide a good standard follow-up. We also believe this causes our families to be unfairly divided, so those are good health insurance got to follow-up and those without good insurance or with no insurance really didn't get to how that the same follow-up care, and so we question whether this was discriminatory. Next slide, please.

To ensure that all newborns received follow-up care regardless of their insurance status, the newborn screening program developed a contract with our testing lab in Hawaii so that all the thalassemia testing lab will receive all of the samples, alpha thalassemia samples from the Hemoglobinopathy clinic, and so that guaranteed them samples and in return they would provide us with the discounted rate for our testing. The newborn screening program covers DNA testing for alpha thalassemia for the newborn and for both parents, as well as the CBC for all three individuals. We can check the [NCD 00:34:29] and other CBC indices against the result to make sure it makes sense, and it covers a blood draw fee for all three. This arrangement it's also retroactive, so it applies to anybody with a positive newborn screening for alpha thalassemia in Hawaii. Like somebody who came to our clinic as 18-year-old, we still take advantage of this if their Hawaii newborn screen result was positive for alpha thalassemia. Obviously, this really increased the number of families who completed the testing. Next slide, please.

How does our clinic work? Once the newborn screening program gets a positive result for Hemoglobinopathy, they send the contact information for the clinic to the appropriate pediatrician and suggest some referrals to their families. We usually have about one to two and half days of Hemoglobinopathy clinic per month and we schedule about six families per visit and it usually always full. During the initial visit, the family meets with the genetic counselor who provide the education and counseling about the suspected hemoglobin disorder and offers confirmatory testing. The medical geneticist then comes in and does a pretty brief physical exam really just looking for any signs of hemolytic anemia like jaundice or splenomegaly, and [she just basically 00:35:41] clean up, so she answers any remaining questions.

The family then gets their testing done and a few weeks later the counselor calls the family to disclose the results. During the call that counselor also just going to review the information we talked about during the initial visit like the management and recurrence. The follow-up call just helps cement the information discussed for the families. This process is quite really well for us and for many of our families. However, some families were still running into issues. These are mainly our neighbor island valleys. Next slide, please.

This is the short geography lesson for those of you have never had the pleasure of visiting my home state. Hawaii is made up of eight main islands and all genetic services including the hemoglobin clinic are located on the island of Oahu in Honolulu and that's where the red star is. We don't have bridges connecting the islands. We don't have ferries that go between the islands. The only way to get from a neighbor island to Oahu for services is to fly. Now, the average round trip ticket from a neighbor island to Honolulu is about $200 a person and that doesn't include any other travel expenses that a family might incur like renting cars or parking fees.

Some insurance companies they will pay for a child and one parent to fly to Honolulu for a specialty appointment but not all well. We also do organized monthly outreach clinics to the different neighbor islands, but [spots 00:37:08] for these clinics we try to reserve those for kids who really have medical needs as well difficulty travelling. The waitlist for these clinics are also pretty long, can be as long as six months.

As you can see, the geography of Hawaii plus the lack of affordable transportation between the islands really creates a barrier to accessing services. For neighbor island families who just have a newborn screening result, positive result for alpha thalassemia, not all of them want to make that trip to Honolulu for appointments. Next slide, please.

With these issues in mind, we created a telemedicine program in June of 2006 to five neighbor island sites, the island of Kauai, Molokai, and Maui and two sites on the big island of Hawaii, Hilo and Kona. Neighbor island families who are referred to our hemoglobin clinic are offered either a telemedicine appointment or in-person or Oahu appointment and they can just choose which they prefer. Next slide, please.

A little bit about our telemedicine program, we use real time video conferencing equipment so the genetic counselor and geneticist and family can see and speak to each other. The interaction is similar to one that you would use if you were doing like Skype or FaceTime. The connection is secure. Next slide, please.

The neighbor island sites are located at our Department of Health offices on the different islands. We also have an onsite facilitator who's usually a Department of Health social worker or public health nurse who sits with the family during the session. They not only work the equipment but they also provide a physical professional presence for the family. We found that this is really helpful to have a clinical professional and not just an IT person present. Mainly just because they can help likely to physical queues for the family, they can help create and make the family feel welcome, and they can also help us coordinate any testing or follow-up for the family. What I like best is we can send them any paperwork like our test requisition forms or anything like that via fax or email. Next slide, please.

This is just an example of one of our educational is some of our families English is their second language that's really helpful especially during a telemedicine appointment when you're not in person with them to have something that they can look at as you're talking and going over the different [inaudible 00:39:31] with alpha thalassemia. Next slide, please.

This is a test requisition form and this is what we can also send to our Department of Health social worker or public health nurse to help us, again, hold any testing. Next slide, please.

The Honolulu site is currently located at our children's center, Kapiolani Medical Center, and right now a medical geneticist and a genetic counselor attend each session. However, we're probably going to be changing this soon. We recently installed brand new telemedicine equipment in our Honolulu Department of Health office, so we're going to be switching our Honolulu site and I think this will make scheduling the telemedicine session easier for us. We're also going to only be staffing the sessions now with just a genetic counselor. Since the Hemoglobinopathy clinic appointments are really just education and counseling and since really a physical exam can't be performed via telemedicine, we decided the medical geneticist time would be better spent doing other things like seeing in-person patients and just revealing and signing off on the genetic counselor session. Next slide, please.

We do use a bridge between the neighbor island and Honolulu site and that just helps the connection work better. It's provided for us by the University of Hawaii through STAN which stands for the State Telehealth Access Network and it just a federally-funded program. They usually charge $36 an hour to provide a [inaudible 00:41:00] service but they waive that for us because we're providing clinical services to the neighbor islands. Next slide, please.

We do satisfaction surveys with our telemedicine clinic to make sure that everything is working well, and it's completed by our family specialists, onsite facilitators and the referring provider and the results are overwhelmingly positive. Families prefer telemedicine to fly to Honolulu or participating in a phone counsel. The families who participate state that they are very comfortable with the method of communication and that the use of telemedicine doesn't interfere with their ability to have a good conversations with the specialist. Since we were worried that using equipment will make families feel like they couldn't talked or couldn't be heard by the specialist, so that was good to hear. Family support they are very confident in the quality of care provided. The geneticist family, onsite facilitator, and referring provider all state that they would strongly recommend telemedicine to other families. Next slide, please.

As far as reimbursement, we've been tracking reimbursement for telemedicine since the beginning. The medical geneticist submits a claim using a modifier code to indicate that services were provided via telemedicine about makes

tracking really easy. We compared it to our in-person hemoglobinopathy clinic visits and we realized that reimbursement improved over time. Usually, it was denied at the beginning but by about 2009 it was being reimbursed at an equivalent rate. I think ensuring companies just needed time to educate themselves about telemedicine. In 2009, Hawaii passed state legislation about telehealth reimbursement. Next slide, please.

We've been really lucky that other class often associated with telemedicine clinics to apply to us because of our really great private-public partnerships. I think buying charges are waived by the University of Hawaii and the Department of Health. The room rental or facility fees also don't apply to us because the rooms are provided by Kapiolani Medical Centers and to the Department of Health in Honolulu as well as the Department of Health on neighbor islands. I apologize that last bullet is incorrect. It should say Kapiolani Medical Center instead of University of Hawaii. Next Slide.

However, if the cost that were waged for us were included, our current rate of reimbursement for telemedicine at least wouldn't cover the cost of our telemedicine clinic. Instead, to make our telemedicine clinic sustainable, we've developed really, again, great strong public-private partnerships. Actually, these partnerships that helped to make not just the telemedicine clinics successful but also our Hemoglobinopathy clinic to success and really also allowed us to provide confirmatory testing to all family to the positive newborn screening result for alpha thalassemia. Our suggestion to other states speaking about beginning a similar program for newborn screening conditions is to just look at those around you and figure out how you can develop partnerships or collaborations that can bring you closer to your goals of providing the best care for your kiddos. Next slide.

That's the end. Thank you for listening to me and I'm happy to take any questions.

Sikha: Lianne, that was wonderful and so insightful. We do have one question that came in via the chat which hopefully you can answer prior to taking phone questions. That question was, are there any forms of Hemoglobinopathies where you would recommend an in-person clinical visit rather than the telemedicine video conference?

Lianne Hasegawa-Evans: That's a good question. Even for our probable hemoglobin H disease we have done it through telemedicine, however, with the caveat that those kids get either a pediatric hematology or a pediatric geneticist to consult at some point before their first year. They don't have to fly out just for the genetic counseling appointment, but we do again recommend that they come out. We don't see a lot of sickle cell trait or sickle cell disease but I think the same thing would apply if we did get a positive result for sickle cell disease. We probably want those kids check out at least at some point by either medical geneticist or a hematologist,

because, again, that physical as I'm looking for hemoglobin H disease, looking for hemolytic anemia is pretty important.

Sikha: Thank you. There is another question that just came in asking how does this work with HIPAA's laws? For example, providing results over the phone and not knowing who is on the other end of the line. Are there any HIPAA-related implications for that?

Lianne Hasegawa-Evans: I think that's referring to after this in-person visit or after the telemedicine visits. We do meet our families either be a telemedicine or in-person to give them free counseling information about the task and as well about as well as their newborn screening result, then we give the test requisition form at that appointment, they get the testing and then we disclose results over the phone after that. Usually it's pretty I think … because we'd have that first visit with the family and we know who to disclose it to. Since most of the time the newborn screening result is pretty accurate so we do counsel as if that result is correct given the caveat that it's still screening program and if that confirmatory testing is necessary most of the families understand and I think it's fine because, again, we have that initial visit either be a telemedicine or in-person [inaudible 00:46:50].

Sikha: Okay. There's no additional questions in the chat box, but if there's any questions on the phone please hit star seven to unmute your line.

All right. John, we're not hearing any additional question. Would you want to say anything to wrap up today?

John: Yeah. I like to thank the presenters, thank [FHL 00:47:20] for supporting our efforts here at the short term follow-up workgroup. Once again, if you have … either if you have further questions you can send an email to Thalia, the email address is here. We'll send you a short survey. Our workgroup really takes careful look at what you say in those and hopefully you'll see that we're responding to some of your comments in the different efforts that we are making as a group. Once again, this webinar was recorded, so if you found yourself thinking, "Oh boy, I wish that one of my colleagues were here or have seen this," you can send out the links to those people that would benefit from it when we have that available. It's usually within a week or so. We'll send out through the LISTSERV. Thanks so much once again to Sharon, Felicia and Lianne.

Sikha: Thank you.

John: Bye everybody.