NewSTEPs
A Program of the Association of Public Health Laboratories™

Short Term Follow Up Technical Assistance Webinar

September 2014

Presentations:

- State Profile Alaska—Marcy Custer, RN, MS
- Genetics Counselors Enhancing Newborn Screening Activities—Sylvia Mann, MS, CGC
- The Role of a Clinical Genetic Counselor in Newborn Screening—Andrea Atherton, MS,CGC

Moderator: John Thompson, Co-chair, 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, [inaudible 00:05] to everyone. I'm going to push the Mute All button now so you will be muted.

Speaker 2: The conference has been muted.

Thalia Wood: I would like to remind those of you who are speakers today just to push Star 7 to un-mute yourself and then you can push Star 6 to re-mute yourself once you're done speaking. John, are you on the call?

John: I am.

Thalia Wood: Okay, you want to go ahead and get us started John, that would be great.

John: Thank you, Thalia. Well, I'd like to welcome everybody on behalf of the Short Term Follow-up Committee who plans these calls to this technical assistance webinar. I will be talking about genetic counseling and newborn screening. First off the agenda. What we'll plan on hearing, who will be speaking. First we'll be hearing from Marcy Custer from the Alaska Newborn Screening program. She's going to give us an update from Alaska.

She'll be followed by Sylvia Mann who is in Hawaii. She's a [state 01:12] genetic counselor there. Then our final speaker will be Andrea Atherton, from Children's Mercy Hospital in Kansas City.

They'll be each be speaking about their unique roles in newborn screening as genetic counselors. We're excited to hear from them.

Before we get to their slides I'd like to give you a couple of teasers for the upcoming newborn screening genetic testing symposium that's happening October 27th through the 30th in Anaheim, California. Coming right up. First of all Carol Johnson submitted an abstract from Iowa's newborn screening program. She and I jointly submitted a round-table and we hope to invite ... hope that many of you will able to come to that round-table. We'll be talking about short-term follow-up and some of the challenges therein.

Then, at the symposium also, so next slide, there we are. We're going to have a short-term follow-up mixer. This is a chance on Monday, the first day of the symposium for us to get together after the opening social to have our own opportunity to meet with each other. We know a lot of each other by face, but many of us are new or haven't had the chance to come to these types of meetings so this will be a chance for us to meet face-to-face and talk about some of the challenges that we have.

We'll also encourage you to ... we're going to have on display educational materials from the different programs so if you could choose two or three of the favorites, things that you're especially proud of or that may be unique to your program to share. It can be a printed copy or we'll have availability if you want to bring a laptop and have it hooked up that way.

I wanted to give you a special welcome to both of those activities, focused on short-term follow-up and outside of the normal program, which does include a couple of breakout sessions of follow-up too.

We're very glad to have that and our committee's been working hard to prepare good activities. We hope that you come to that. I think that Thalia has a couple of things to stay from [inaudible 03:53] and we'll get on to our presentation.

Thalia Wood: Thank you very much John. I just wanted to point out to everyone. I hope you like the NewSTEPs.org website. I just wanted to remind you that you must be on the listserve, you probably are since you found about this webinar, but there are other listserves, like the Health Information Technology listserve and we also have LinkedIn.

We often announce these webinars and pertinent information about newborn screenings through LinkedIn so feel free to join our LinkedIn group as well. Again, don't forget you can always send me suggestions for future calls and there will be a short survey sent out after today's call as well.

We'll go ahead and get started with our state profile. Marcy go ahead and do Star 7 and just tell me when you want me to advance the slides, Marcy.

Marcie Custer: Thank you Thalia. This is Marcy Custer. I'm the perinatal nurse consultant with the Alaskan Newborn Metabolic Screening program. Can you go ahead and advance the slide.

I thought I would give a profile of Alaska in relation to the recommendation around timeliness of newborn screening. In particular that the specimens be received at the lab within 24 hours. Advance please.

I thought it would be good to have a look at births in Alaska. This is a map of our fertility rates. You can see that with western and northern Alaska the areas are dark. Those are areas that we have a lot of births and these are also rural areas of our state. Since 2006 we had nearly 11,000 births in our state and since then we've had slightly over 11,000 births annually. Because of these numbers we don't have newborn metabolic screening in our state. We contract with the Oregon Public Health Lab. Next slide.

Another interesting fact is that we have a lot of out-of-hospital births in our state. In March of this year the CDC published a data brief on the trends of out-of-hospital births in the U.S. and in 2012 Alaska had the highest percentage. We were 6%. The other blue states also have a high percentage of out-of-hospital births. This means that we have a lot of midwifery centers in our state and that we have a lot of midwives who are collecting newborn metabolic screening. Next slide.

Who else in our state is collecting newborn metabolic screening? This just tells you a little bit about our health care system in Alaska. We have private hospitals, pediatricians, family practice providers, primary care providers. We have a tribal health system and this is a picture of the Alaskan Native Medical Center in Anchorage. But in some of our rural hub communities like Bethel and Dillingham we also have hospitals in some of those communities.

Then when we branch out to the small villages the tribal system includes village clinics that are staffed by community health aides. We have some military facilities in our state. In Anchorage we have joint base Elmendorf-Richardson and in Fairbanks we have Eielson Air Force Base and Basset Army Hospital. We also have several federally qualified health centers that charge a sliding scale fee to the clients and we have some of these located in our larger cities like Anchorage and Fairbanks, but we also have them in some of our smaller rural communities. Next slide.

This is a photo of the rural community of King Cove and you can see we've got a couple of signs there. We've got one with a bear and then we've got some children on a teeter-totter. The building to the right there, behind the signs, is the school and I'm told that the clinic is just a little bit past this building. I just wanted to give you a snapshot of a rural community. Next slide.

We often hear about using courier services to improve the timeliness of newborn screening. For our state that becomes somewhat of a challenge because of our geography. We are a very large state. We're one-fifth the size of the United States. Next slide.

We have a lot of mountains in our state so we have a limited road system and the red box that I've highlighted there on the map is the main area where we have a road system. We don't really have roads going to western Alaska or out on the Aleutian Chain or anything like that. In southeast Alaska we have islands and of course they have roads on those islands but to travel between islands you would need to travel between the ferry system or travel by air. Next slide.

A lot of travel to rural Alaska is by aircraft. This is just a picture out of a small plane and you can see that we have a lot of mountainous terrain. Next slide.

Let's just look at an example of getting a specimen from Bethel, Alaska which is located in Western Alaska down to the Oregon lab down in Hillsboro. You can see that a jet flight from Bethel to Anchorage is about 400 miles and it takes a little more than an hour. Then a direct flight from Anchorage to Portland is about 1,500 miles and is nearly 4 hours. Then Hillsborough is about 17 miles from Portland so that could take about another half hour depending on traffic. Next slide.

The use of FedEx. We know that some of the facilities in some of the communities that have FedEx do use this service to get their specimens to the lab in a timely manner. But we do not have FedEx in all of our

communities. It is mainly located in urban areas. I checked with FedEx and these are the five communities that they have a shipping center in Alaska. Next slide.

I'd like to share with you the great work that the Yukon Kuskokwim Health Corporation in Bethel is doing to get their specimens to the lab in a timely manner. Just remember they're about 400 miles from Anchorage. They're out here in western Alaska. YKHC is a tribal organization and they serve 58 communities. I like the little map that they have insert there in the lower right corner; you can see the orange area is the area that they serve. I know that the numbers by the communities are small for you to see but the green numbers tell you the air miles from the village to Bethel and then the orange numbers tell you the flight time in a Cessna, which is a small plane, from the village to Bethel.

For example, if we look at Grayling, which if you want to see where that is it's kind of in the north central part of the map there. They are located 146 air miles from Bethel and it takes about 75 minutes on a Cessna to get to Bethel.

The YKHC lab has set up a system where their village clinic collects the newborn metabolic screening, and then they hand-carry that and give it to the pilot or someone on the plane going into Bethel. Then the lab makes rounds to the small air taxis three times a day to pick up the specimens, the screenings. Then they express mail them to the Oregon lab. I thought that was a pretty great system that they've set up. Next slide.

We do still sometimes receive back, lab samples that say it's too old for testing and we do sometimes have transportation delays due to weather in our state. It's not uncommon to have snow storms during the winter or strong winds that might halt air travel.

For me my F word is fog. My personal experience as an itinerant public health nurse traveling to Dutch Harbor more than once I was delayed three days trying to get in or out of Dutch Harbor because of fog.

We also have volcanoes. We have a chain of volcanoes down on the Aleutian chain in south central Alaska and sometimes volcanoes will spew ash and that will halt all air travel.

Then ice storms can also be a problem that sometimes cause closures. We've had some of that recently in Fairbanks and Anchorage. Even this

last year the Oregon lab had some closures due to ice storms down there. Next slide.

Our most common findings on newborn metabolic screening is the CPT1A Arctic Variance. During the past year the cut-off for the first sample has been lowered and so this has really helped with identifying some babies much earlier. This slide is courtesy of Dave Cohler. He's our consultant for the Alaskan newborn metabolic screening from the Oregon University. This just shows you a distribution of where we have children identified with CPT1A.

We've had a collaboration to produce an educational DVD and this was a partnership with the Alaskan Native Medical Center, the Norton Sound Health Corporation in Nome and the State of Alaska. We send out the educational DVD to parents when an infant is identified with CPT1A and a letter goes along with that signed by Matt Hirschfield. Dr. Hirschfield's from the Alaska Native Medical Center and Meg Kurtagh with the Alaska Newborn Metabolic Screening program. Next slide.

I'm the one that's involved with the follow-up calls and I have made contact with healthcare providers in all of the different healthcare systems that you see listed here and that we've already discussed. Next slide.

Also, for quality assurance, we sometimes receive lab results that are unsatisfactory for testing, such as not having enough blood or uneven saturation or contamination, so for quality assurance we address this by giving QA feedback reports to providers. For this fiscal year we've also budgeted some money for travel to do some sight test visits. Recently we've collaborated with Providence Hospital in Anchorage to produce a short 6-minute training video, so we now have that available on DVD to use with our providers and we'll also be making that available online. Next slide.

In closing, these are our Alaska babies. We're all in this together. Our healthcare system involves many partners. We have healthcare providers across the state involved in collecting specimens. We are a large, vast state. We have geographical barrier with long distances and beautiful mountains but this has created a limited road system for us so air travel is very common. We have a little more than 11,000 births per year so it's not really feasible for Alaska to do newborn screening within our state and this is why we contract with the Oregon lab. And that's it.

Thalia Wood: Great, thank you so much Marcy. We appreciate that. We'll move it right on into the next presentation so we want to make sure everybody had a chance to ask questions at the end and have discussions.

Our next presenter is Sylvia Mann from the State of Hawaii. Go ahead and do Star 7. Are you un-muted Sylvia?

Sylvia, Star 7 to un-mute your phone.

Sylvia Manns: Okay.

Thalia Wood: Oh, there we are. Okay thanks Sylvia, go ahead and tell me when you want me to go ahead with the slides.

Sylvia Manns: Great. Hello everyone. I guess Thalia asked me to talk mainly because we have a lot of genetic counselors that work in our program. I'm going to talk about how genetic counselors enhance our newborn screening activities. Next slide, Thalia.

I thought that I better start by telling you how our health department is structured and where the genetic counselors are. Back in the early '90s the administration of the health department was smart I suppose, and decided that they wanted to have a separate genetics program and have a state genetics coordinator. That's when I was hired. I am a genetic counselor by training but I was not hired within the health department to do genetic counseling.

At that point there was a genetics program and in about 2012 the health department did a reorganization and instead decided that they were going to consolidate a bunch of programs under the genomics section. The genomics section is under the children with special health needs branch which is one of our Title V agencies, for those of you who work in maternal child health and work with block grants, you'll know what Title V agencies are.

We have a genomics section and under the genomics section we have a birth defects program. We have the genetics program. We have the newborn hearing screening program and the newborn metabolic screening program.

I am supervisor of the genomic section. Next slide.

For our newborn hearing screening program we have a fairly small staff. It's a program coordinator, a social work assistant and an office assistant.

Then we have a [Hirsa 18:34] called Baby Hears to improve our newborn hearing screening, so we have a grant coordinator and a project specialist. Next slide.

For our newborn metabolic screening program we have a program coordinator, follow-up nurse, med-lab tech and an office assistant. The staffs are small in our metabolic screening program and our hearing screening program.

Our laboratory services, like Alaska, are contracted to the Oregon Public State Health Lab. We have about 19,000 births a year and it definitely is much more cost effective for us to contract with Oregon than to do the lab testing within the state. We are lucky in that we do have FedEx that goes to all our islands. Our samples, lots of times get to the Oregon State Lab before Oregon baby samples get there. We had really no problem with the timeliness article, thank goodness. Next slide, Thalia.

We're the genetic counselors in our health department. As I said I'm a genetic counselor but I don't do any genetic counseling. Within our genetics program we have three genetics counselors, LeeAnn Huskaow, Kristy McWalter, and Jennifer Boomsmall. All of them split their time between doing public health genetics work and doing clinical counseling. I'll explain that a little later on about the clinical counseling and how we collaborate with our clinical unit. Next slide.

I figured I'd break down some of the roles that the genetic counselors have in what we do for our newborn screening activities. The good part is because we have a separate genetics program and other staff; our newborn screening staff is able to concentrate on our newborn screening program. On the day to day operation of the program and on making sure that the kids are screened, the personnel in the hospitals are trained, and that we can do short term and long-term follow up in the newborn screening program with the newborn screening staff.

Genetic counselors help with our newborn screening activities when we do policy. We just finished convening our SKID task force. When we do add disorders to a newborn screening panel the genetic counselors help by providing research support for the task force meeting, drafting protocols, drafting information education materials that might be needed and even helping convene the task force.

The genetic counselors also help sometimes draft, track and work on newborn screening legislation so that the newborn screening staff can

concentrate on doing what they're supposed to be doing, newborn screening. Next slide.

This is one of the education brochures that we developed around CCHD screening when we had a CCHD task force. It's very nice that we could have the genetic counselors develop things like these education materials when we're having discussions on adding disorders. Next slide.

The next thing that we do a lot of is community engagement. Genetic counselors are great at community engagement. We have done interviews, focus groups, surveys and most of these community engagement activities are around community attitudes towards newborn screening. How do they want disorders added? How do they feel about follow-up? What do they want for services? How do they want their educational material delivered to them? Genetic counselors do a lot of community engagement. Next slide.

This is one of our activities. We have found that baby expos are great places to get community engagement. People are either planning babies, pregnant already or just had babies can come over. This one is actually an iPad enabled survey that we were doing on the audio system to get some community perspective on newborn screening. Next slide.

Of course one of the big things that genetic counselors help with is education, in developing brochures, resource guides. Right now we're doing parent resource guide for newborn hearing screening; fact sheets, website content and of course doing presentations both locally and nationally on our newborn screening activities. Next slide.

One of the big educational activities that we've done over the past decade is that genetic counselors have helped with our newbornscreening.info site. This is a site that has our parent fact sheets for each of the disorders that are screened. I think on the last count there were 38 fact sheets on this site. Next slide.

One of the fact sheets is on PKU1. It basically has information that parents said that they wanted to have when they first got the phone call of the diagnosis, while they're waiting to go see the specialist to be able to find out exactly what was going on. We also have found that these fact sheets are very useful for the primary care providers who like to use them to use as a cheat sheet before they actually phone a family to tell them that they have a positive newborn screening result.

There will actually be a talk at the APHL meeting about our decade of experience with these fact sheets. Next slide.

The other thing that we have the luxury of doing is doing research. The genetic counselors help develop research ideas, help obtain extra funding, actually do the research. We generally use the findings for our program because that's usually why we're doing the research and we publish to help other people use the research. We also go and present findings outside the program. A lot of our research is around the ethical, social legal issues with newborn screening.

We also do research when we're trying to figure out if we need to follow some national guidelines. When the NICU guidelines were put out for a newborn screening we actually did a research project with our largest birthing hospital to find out if the guidelines helped our NICU babies or had no effect or had some adverse effect on our NICU babies. We actually found, after doing 6 months of following the guidelines, that compared to previously when we weren't following the guidelines we didn't miss any more babies or catch any more babies. Instead the babies in NICU were getting stuck a lot more than they did previously of course.

The NICU perinatologist and the program decided that probably with a small state with a very tightly run newborn screening program, with babies that do not get discharged early; we hardly have any babies discharged before 24 hours in our state. Probably following the NICU guidelines really just resulted in the baby getting more heel sticks than necessary. At least we had the evidence to show that following the guidelines really didn't help our state too much or at least our NICUs in our state too much. Next slide.

In some of the publications we did some ethical, legal, social issues research around expansion of newborn screening. We've helped Guam with some of the needs assessment for their newborn screening. Next slide.

So for our clinical role I know Angie is going to talk more about clinical counseling. For Hawaii we have a collaboration for our clinical genetics with our children's hospital Kapiolani Medical Center, so we have a collaborative clinical services where everybody puts in resources so we can maintain our pediatric genetic services.

The partners are the children's hospital, Queens Medical Center, Department of Health, and the University Of Hawaii Medical School. Everybody puts in resources to make sure that we do not lose our

pediatric genetic services which we did about a decade ago. We lost it for about two years and that was very, very bad. We never want that to happen again. Our genetic counselors will sometimes see the metabolic patients in clinic when they need genetic counseling. For routine visits, genetic counselors aren't involved in those visits.

We also have a [inaudible 28:06] clinic where the genetic counselors are involved and we also do tele-genetics. For tele-genetics we mostly do that counseling for alpha thalassemia, because in our state we find about 35 carriers in the newborn screening per month.

The family is offered counseling and also we do mutation analysis because we need to figure out what mutations are actually going on in that family and whether the family has hemoglobin H or two mutations, one mutation. Part of our newborn screening follow-up is we actually do pay for the DNA mutation analysis for the mother, father and the child to be able to do the genetic counseling. That has been done through tele-genetics and in person at our hemoglobin [inaudible 28:58] clinic.

Starting at the beginning of next year I'm told Oregon will start doing RRT DNA for cystic fibrosis screening. Right now they are doing RRT/RRT. So we will be finding carriers and one of the things we're planning is to also do cystic fibrosis carrier counseling through tele-genetics.

Luckily we're in Hawaii which means our Caucasian population is very low. We really don't have too many cystic fibrosis cases that are diagnosed which means we definitely don't have that many cystic fibrosis carriers diagnosed either. This should hopefully lend itself well to tele-genetics. Next slide.

I thought at the end I would talk a little bit about the skill set and training of genetic counselors. I find them well-suited for newborn screening activities. I actually find them suited well for all our activities, mainly because they're trained to take very complex concepts and develop methods to explain them in plain language, which means that they can talk to a wide-range of stakeholders; from families to legislators to policymakers to our other program colleagues in the health department and our providers too.

They have an understanding of both the clinical and psycho-social aspects surrounding newborn screening so that it helps in developing our education material, talking to stakeholders and developing activities. As I said, genetic counselors are great at developing education materials so definitively an advantage for newborn screening.

Then on top of it genetic counselors can provide clinical services to families. For families that need genetic counseling services if you have a genetic counselor on staff you can do things like the tele-genetics. You can do things like having a second hemoglobinopathy clinic that's run out of the health department. Next slide.

That's an overview of what we do in the health department with our genetic counselors. We are very fortunate to have genetic counselors working for me and I am very lucky to have very, very dedicated and wonderful counselors that work in the program and are willing to do all this work that they probably thought they would do in their life.

Thank you.

Thalia Wood: Thank you Sylvia. That was a wonderful overview of the whole Hawaii program.

Up next we have Andrea Atherton from, I think you're in Missouri, is that correct Andrea?

Are you in Kansas City ... don't forget to do Star 7 Andrea to un-mute yourself.

Andrea?

Andrea Atherton: Are you there?

Thalia Wood: Andrea, is that you?

Andrea Atherton: Yes.

Thalia Wood: Yes, we can hear you now.

Andrea Atherton: I have two mutes on my phone.

Thalia Wood: Oh, no problem. Thank you. You're in Kansas City, Missouri, is that correct?

Andrea Atherton: I am in Kansas City, Missouri.

Thalia Wood: Thank you so much. Go ahead and get started.

Andrea Atherton: No problem.

My role is I am a clinical genetic counselor. I am employed by Children's Mercy Hospital and my primary role is in medical genetic counseling.

In hearing all the conversations and presentations before I'm very jealous because I wish we had genetic counselors who worked directly in our newborn screening program to develop the resources that other program have had the ability to develop. As a clinical genetic counselor we actually use a lot of the resources that came out of the Hawaii program for our patient population. There's no sense in re-inventing the wheel if somebody has fabulously done it in the first place. Next slide.

My primary role at Children's Mercy is I'm a full time clinical metabolic genetic counselor in an independent free-standing children's hospital. I primarily see patients with inborn errors in metabolism, [inaudible 33:18] storage disorders. Any newborn screening that comes through I'm the primary point of contact for any abnormal newborn screen in the state of Missouri, the western third as well as the entire state of Kansas.

I also see and help to work up patients with neurological and neuromuscular disorders with our rehab medicine and neurology group here at the hospital. Next slide.

I'm going to talk a little bit about the genetic services in Missouri and then briefly go over genetic services in Kansas since we're the referral center for the entire state of Kansas as well.

In the state of Missouri, Missouri newborn screen actually contracts with four different clinical sites hospital in the state of Missouri that any abnormal newborn screen that comes through the newborn screening lag gets referred to a genetic counselor or metabolic geneticists for follow-up.

We provide clinical genetic services in Missouri including follow-up services for newborn screening. We [inaudible 34:24] contract states with each site so each must have one clinical or medical geneticist on staff that is trained in syndrome identification. The contracts that we have from the State of Missouri for genetic services state that we have to have a provider on staff. But it also states that the support that the State of Missouri gives us for genetic services can also help to pay for salaries for other individuals on our team such as genetic counselors, biochemical geneticists, nurses, clerical staff, etcetera.

The clinical sites are responsible for any newborn screen that is referred. If we get a referral we are the ones that are in charge of making sure that

family is notified, the primary care physician is notified and that the appropriate follow-up steps are then taken, are completed to either get that baby diagnosed on treatment or closed out as false-positive. Next slide.

This is just a pictograph of the State of Missouri. Annually our birthrate is around 80,000 per year. We're divided into three regions, kind of straight down the middle. We have a western, which I'm in Region I at Children's Mercy Hospital, pointing where the arrow is. The middle section of the state gets referred to Columbia, Missouri, University Hospitals and Clinics and they're located pretty much in the center of the state. Then Region III has two clinical referral centers at St. Louis Children's and at Cardinal Glennon and Children's. The referrals are based up based on baby's last name, or all of the babies that are born at St. Mary's get referred to Cardinal Glennon. Next slide.

Genetic services in Kansas is a little bit different. As I stated, in Missouri any referral that we get on babies that screen positive come directly to us and we're responsible for making sure that gets followed up. In Kansas the way the law is written is that all of the newborn screens that are referred as abnormal go back to the provider of record on the newborn screen and that provider is actually the one that's responsible for following up on that abnormal newborn screen.

Kansas is ... we recently entered into a contact with Kansas in the last year to help provide genetic services for abnormal newborn screens so that the primary care physicians on record can have a place to call with questions or concerns on the newborn screens that they get.

We also get copied on any abnormal newborn screen that comes through in the state of Kansas. We get both an email and a fax from the state lab in Kansas, so if there's any newborn screen that looks suspicious to us in which follow-up needs to be coordinated in a timely manner I reach out to the provider of record to make sure that they have all the resources available to them to get the correct labs on the baby, or even offer to see that bay in our clinic within 24 hours. Next slide.

For us, in a clinical position, newborn screening is completed in the state lab in Kansas or Missouri. For me in Missouri the abnormal test result is sent to the appropriate referral site.

Specifically for us we get all of the inborn error of metabolism referrals. We are also doing newborn screening for [inaudible 37:57] storage disorders in the state of Missouri so those referrals come directly to me.

Any referral for an endocrinopathy goes to an endocrine nurse here in the hospital. The hemoglobinopathies get referred to hematology. Cystic fibrosis referrals get referred to our nurse coordinator in pulmonology and our hearing is either referred to audiology or ENT.

We're a big group within one hospital that follows up on a small number of babies and we all kind of work independently on our own little area. Next slide.

I wanted to capture kind of a day in the life of a metabolic genetic counselor that is the consultant or the contractor for providing genetic services to babies, picked up [38:45] newborn screening.

When a newborn screen is referred to the appropriate clinical site we have clerical staff here that logs our newborn screen, refers that screen to our clinic nurses and they create a medical number for the patient. The clerical staff then scans the newborn screening report into the medical record and then that scanned report is automatically given to me.

What I do once I get an abnormal newborn screen is I contact the primary care physician that's listed on the newborn screening report and if the primary care physician on the record is not the baby's actual primary care physician I have to backtrack and try and figure out who the baby's actual primary care physician is, and it's usually by talking to medical record departments at the birth hospital.

Most of the time the family has listed a physician that they were going to be following up with once discharged from the hospital, but not always. Next slide.

Once I identify who the primary care physician is I typically discuss the newborn screening results. If they did not receive a copy of the newborn screen I forward that copy to them and then we decide between the two of us, the PCP and myself as a counselor who is going to be the one that calls the family.

A good majority of the time the primary care physicians default that to me so then I'm calling these families, but I usually like to have the primary care physician call the family to let them know that there was an abnormal newborn screen and that somebody from Children's Mercy is going to be calling them.

In the event that we can't figure out who the primary care physician is I usually end up having to call the family directly and explaining what's

going on and that's a difficult phone call to make to a family that I don't know, because there can be some mistrust. A lot of no-answered telephone calls, a lot of screening telephone calls and back and forth to figure things out.

Once we identify the family and get them on the phone we obtain who their primary care physician is. We clinically assess the baby over the phone. We make recommendations for labs and an appointment and then we call the primary care physician after that telephone call to update them on the baby that's going to be entering their practice and get them all up to date on this family. Next slide.

When the family comes in the genetic counselor will see the baby with the biochemical geneticist and if a metabolic dietician needs to be involved in the first visit, depending on what diagnosis the baby screened positive for, we kind of go in as a team. As a genetic counselor we do the typical genetic counseling thing such as obtaining a prenatal birth and family history, educating families on newborn screening and the condition the baby screened positive for.

We get them educational resources regarding the condition screened positive for and we serve as the spoke of the wheel for this family as the point of contact between the physician, the lab, sometimes the dietician. But if the baby has diagnosed with a metabolic disorder in which the primary point of contact is the metabolic dietician the genetic counselors hand over the torch to the metabolic dietician.

Primarily we are only involved in seeing babies in the clinic while they're being worked up for their diagnosis and immediately when they are diagnosed and all of the genetic testing has been completed for education purposes.

In the future we're involved depending on the complexity of the disease, how often they're seen in clinic. If they're not seen very often or if we're asked to come into the room for family planning we see families back from time to time for genetic counseling. Next slide.

I think I spoke about that. That's ... if a baby has an inborn error in metabolism that requires dietary intervention the metabolic dietician becomes the point of contact for those families, and if the baby does not have an inborn error that requires dietary intervention the genetic counselor is the point of contact for that family until either confirmed or proven as a false positive. Next slide.

Some of the dilemmas that I encounter on the clinical side are that often the pediatrician on record that signs off on the newborn screen is not the baby's actual pediatrician, so there can be some lag in time in trying to identify who's the correct doctor to call and refer these newborn screens to.

Oftentimes the baby's last name has changed, and we're never provided with the baby's first name. Sometimes babies have been adopted out, names changed and there's another issue with trying to figure out where that baby has gone. We had a baby born last year to a surrogate mother so trying to track down the biological family was somewhat of a dilemma.

Then sometimes we get newborn screens referred to us in which a phone number for the parent has not been entered in or it was entered incorrectly. The address is incorrect, mom's name is incorrect and so those can all be factors that may not seem very important to put it on a newborn screen but it is very important if the newborn screen is referred so we can get in contact with the family in a timely manner. Next slide.

This is kind of a joke in our world that you can add being a private detective to another list of duties as a metabolic genetic counselor because we do spend a lot of time behind the scenes just trying to find things out about babies. Next slide.

Contracting with newborn screening labs from a clinical and newborn screening perspective that we have to work symbiotically together to improve newborn screening in general. The personnel in our newborn screening labs are highly trained scientists and are leaders in public health, but to the best o my knowledge there's not a genetic counselor that works directly in these programs. It's essential that there's effective communication between the newborn screening lab as well as the clinical teams to improve newborn screening.

What our states do is we have biannual meetings for a genetics advisory committee that the provider is not just those in inborn errors in metabolism but those in hearing, cystic fibrosis, hemoglobinopathies, endocrinopathies, everything that we screen for come together twice a year to discuss issues related to the newborn screening and how we can improve things.

We also work on different standing committees or task force as new conditions are added to the risk panel to strategically and logically get these conditions added in a timely and good manner. But there's many different groups that occur for task force and committee that are not

specific to inborn errors in metabolism so this supersedes even the little part of newborn screening that I provide clinically. Next slide.

Some of the pros that I was thinking about while putting this slide together being a clinical genetic counselor is that I get to work closely with newborn screening labs to ensure timely diagnosis and the follow-up of patients. I'm the one that's helping to see these patients first-hand and follow them clinically, sometimes for a really long time. Because most of our patients are fairly complaint.

While [culling 46:24] out abnormal newborn screen results can be quite difficult. I can help control the flow of information that is provided to the primary care physicians and families so that this isn't a scary event that's happening in their life; that they're given good information from somebody who really understands this that can hold their hand through this so it's not so scary.

I can help with the clinical assessments and provide education first-hand to families, and do long-term follow-up. Once it's closed out from a short-term follow-up we get to see these babies grow into adults and have children of their own at some point in time. Another pro of this is that many of the conditions that we screen for in newborn screening have treatments so we're seeing improved outcomes long-term and we develop a relationship and rapport with these families, sometimes to the degree of being an extra family member to these families.

We also get to serve as a point of contact between family physicians and the newborn screening lab as well as all of the other things that are needed in the routine care of babies and children that have metabolic diseases. Next slide.

In trying to determine the cons of being a clinical genetic counselor outside of the newborn screening program it was hard for me to think of a lot because I've never worked directly in a newborn screening program. That was one of the technical limitations is not being aware of all the inner workings and technical details that go into newborn screening and general performing newborn screening and all of the policy that needs to be done to get to a point of being able to screen newborns for genetic conditions and get those referred on.

Some of the other things that I've not been directly involved in is the improvement of newborn screening as well as some of the other concerns can be timing or ability to do provider education on newborn screening and disease awareness. Being clinical I'm in the clinic a good

majority of the time and that does not allow me any, or a lot of time to do anything outside of my clinical duties.

While communication is good between the newborn screening lab and the clinical providers there's a time delay in getting results to the correct provider if you're working with contract providers as opposed to having a genetic counselor and [inaudible 48:51] in a newborn screening program that cull those results out directly. I'm another line in that telephone communication to provide communication to primary care doctors. Next slide.

That's the end of my talk. Thank you for having me today.

Thalia Wood: Thank you so much Andrea, that was a wonderful presentation. We actually do have one question already that was typed in. Do you ever use tele-health for that second visit after test results are back?

Andrea Atherton: We are just now gearing up to start a tele-medicine clinic in October. Geographically we pull from about a 3-hour radius in any direction coming from us. Some of our more rural Kansa families may have up to a 7-hour drive so we're not set up in Southwestern Kansas to provide tele-medicine but it is moving in that direction.

One of the difficulties we've encountered as genetic counselors is billing in our state in which there's no licensure for genetic counseling. I'm not sure that licensure would help with billing in this instance because we are able to bill genetic counselor fees at Children's Mercy Hospital, but we do it as a facility fee. One of the barriers we're finding is that if we see a patient through tele-medicine and if they don't physically come to our hospital we can't bill a facility fee which means we can't bill for our services.

We're trying to figure out a work-around so genetic counselors can continue to be involved. Otherwise we're going to have to pass the torch to a metabolic physicians that we work with in seeing those patients if driving a distance to our facility is a hindrance for those families.

Thalia Wood: Thank you. We have another question that was typed in and after if I ask this question is people have questions don't forget to do Star 7 to un-mute your phones.

The next question that was typed in was, can you clarify what you mean by clinical assessment as a genetic counselor?

Andrea Atherton: We just do some basic questions to the primary care physician if they've seen the baby in the clinic to see how the baby's doing and we relay that information to the physician, the metabolic physician that we work with. A lot of times whenever we get an abnormal newborn screen one of the points I forgot to talk about, is once I get it I review it automatically with the metabolic geneticist that I work with and he'll give me a list of questions to ask or things to focus on when I'm calling those results out either to the primary care physician or the family, so that he can have a better idea of what's going on with that baby in case there's an acuity of needing to get that baby into our clinic.

We try on some of the acute cases, if we need to get that baby in that day, we'll stay late to make sure that that baby's seen and started on an appropriate diet, but if it's not as acute we try to get him in within 24 hours, if there's really no acuity. I can typically work with a primary care physician to get the labs done locally and then follow-up once all is said and done for more education for the family or a discussion of any kind changes that our metabolic physician would need to make moving forward; bring them into clinic for that.

It's a very generic clinical assessment that we do as a genetic counselor, being that we don't have any advanced practice nurses that work with us at this point in time.

Thalia Wood: Okay, thank you so much. Anybody else have any questions don't forget to do Star 7 to un-mute your phone or you can type them into the chat box. Or anybody even want to share their own experiences with their own state in genetic counseling?

Speaker 7: Hi, this is Debbie Freedenberg. In Texas we tried to hire genetic counselors to be part of our newborn screening program which is large and very-well organized, but unfortunately our salaries were public health salaries and we could not manage to attract any genetic counselors, so I was wondering how Sylvia managed to get around that?

Thalia Wood: Sylvia, are you still on the phone?

Sylvia Manns: Yes, I'm on the phone. I had to double, un-mute. What was the question again, Debbie? Sorry.

Speaker 7: We tried to hire genetic counselors to do part of our newborn screening program both in an educator role and considering some other things, but our salaries were public health salaries and we could not attract any

genetic counselors because we weren't competitive with the clinical settings out there and I was wondering how you got around that.

Sylvia Manns: When they hired me actually I had to, we can hire above minimum so there's a whole process where you offer a job and if the person declines it and then the program's allowed to offer above minimum because you can go with what is more closer to market value, so they will put you at a different level of civil service to be closer to what it would be out in the private sector. At least that's how it works in our civil service.

Speaker 7: It would be nice if we had that as well.

Sylvia Manns: And then the other way of doing it is that my councilors are, we have a relationship with our university so through our research corporation at the university we hire genetic counselors through them and the salary's set differently from civil service. But they can work within our health department so we have some different ways of being able to [inaudible 54:58]

Speaker 7: I wish those options were available to me, I'm a little jealous.

Thalia Wood: Thank you. Does anybody else have any questions before we wrap it up here today?

John, do you have a few words to say here at the end for everybody?

John: Yes, thank you so much to our speakers. We're grateful for their different presentations. It was informative and interesting and helpful for us so thank you very much.

Thalia Wood: Thank you everyone. If you have any last minute questions you can feel free to email them to me and I will be sending out a short survey again. Thank you to our speakers. You did a great job. Great topic, thank you everyone.