NewSTEPs

A Program of the Association of Public Health Laboratories

New Conditions Webinar

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Presentations:

Please direct all comments/questions pertaining to this presentation to Guisou Zarbalian at guisou.zarbalian@aphl.org or 240-485-2736

Guisou: Okay Jelili. I think we're good.

Jelili: Thank you.

Good afternoon everyone and thank you for joining us for this webinar. The first of its kind for us as relates to these conditions. We appreciate your time and effort in joining us this afternoon.

I'd like to use this opportunity to also thank the co-chairs of the New Conditions workgroup as well as the members of the New Conditions workgroup for their efforts in bringing this webinar and future activities that we plan to engage with state newborn screening programs in the future.

I'm supposed to give a brief overview of the new conditions work group. How we came to where we are and some of our future kinds of activities which I will discuss later on at the end of the webinar.

I should also like to thank Dr. Caggana for actually accepting our invitation to present on our inaugural webinar here.

As you all know NewSTEPs newborn screening technical assistance and evaluation program is a program that is under APHL is funded to provide a number of things to state newborn screening programs. Technical assistance. Evaluations. Evaluation strategies. The comprehensive resource center. Whether it's on quality indicators or other kinds of technical assistance as relates to the newborn screening system.

We've done this specifically for a number of targeted conditions that have been recently added to the recommended uniforms screening panel. Whether it's congenital heart defect. Critical congenital heart defect. Severe combined immunodeficiency SCID. In that particular instance we worked hand in hand with

the newborn screening translational research network to host a number of these.

It used to be monthly. Then bi-monthly. I think it's now about quarterly webinars. Educational webinars to address a number of initiatives strategies implementation activities.

A means to bring folks and states together to discuss how we can best bring on any one of these new conditions. Whether a state is thinking about it or it's been added to the recommended uniform screening panel at the recommendation of the secretary of health.

We wanted to do something like that for these new conditions that are just been added. As most of you know 3 new conditions were added just in succession starting February of 2015 until last year. PROP and MPS1 and XALD.

Rather than having a specific targeted technical assisting webinar activities for any one of those three conditions at the suggestion of our new [inaudible 00:03:39] committee- Please forgive me there is a fire truck going by here- At their suggestion we decided to change the title. Instead of focusing on MPS1 on the XALD 2 new conditions.

The charge of this New Conditions workgroup as you can imagine is to identify resources. To develop new resources where there are gaps for states as they think and consider. To implement new conditions that have been just added or at least be thought of to be included as part of the recommended uniform screening panel.

Our primary focus will be on those 3 conditions. However we have leeway to focus on new conditions that are being considered to the recommended uniform screening panel. We will bring together experts. Hear from folks in the field. Then newborn screening to discuss these strategies on a bi-monthly basis with you all.

The new conditions work group is tasked with meeting on a monthly basis to form the ideas of whatever is trending that will be of benefit to you all in state newborn screening programs.

We'll stop there. A lot of work has been done over the last several months to get this inaugural webinar going. We look forward to hearing any and all of your ideas on future webinars. On any activities related to any of the new conditions.

I think at this point I'm going to turn it over to Kim Piper and Fizza, if she's on, Majid to give us an outline and introduction of our featured speaker. Kim?

Guisou: Fizza please press star 7 to un-mute your line.

Fizza Majid: I did. Can you hear me?

Guisou: Yes. Okay. Yes. Great. Fantastic. Thank you.

Fizza Majid: Good afternoon everyone. As Jelili mentioned the presentation today is going to be on new presentations for [inaudible 00:06:04] of new conditions by Dr. Caggana. Followed by discussion then later discussion related question and answers.

Now it is my pleasure and an honor to introduce Dr. Michelle Caggana. Dr. Caggana received her doctorate degree from the Harvard school of public health and completed post-doctoral work in clinical molecular genetics at the Mt. Sinai school of medicine.

She is board certified in clinical molecular genetics by the American Board of Medical Genetics and a fellow of the American College of Medical Genetics and Genomics.

Dr. Caggana has been employed by the Wadsworth Center since 1996 where she is deputy director of divisional genetics. Chief of the lab of human genetics and director of newborn screening program. She's the co-chair of the Newborn Screening and Genetics in Public Health Committee for the Association of Public Health Labs.

Dr. Caggana is also consultant to the FDA. Her lab has [inaudible 00:07:07] of several human screening tests and uses DNA technology to study frequencies of specific gene mutations in dried blood spots in the context of newborn screening.

Dr. Caggana?

Dr. Caggana: Thank you Fizza. Thanks everyone out there for calling in. Also to APHL. The New Conditions workgroup for allowing me to speak to you today.

I will tell you that the slides that I'm showing today I actually came up with them while we were at the newborn screening genetics and public health committee at St. Louis. Just prior to symposium this year.

Basically my charge is to give a high level view of all of the different types of things that labs need to consider in order for them to begin implementing new tests. I hope that recycles thoughtful discussion and I welcome questions.

It's my understanding that there will be a follow up on follow up. I will just introduce follow up at the end of this discussion of laboratory considerations. Next slide please.

The first question really is before you start in your own lab you need to consider your state. How are new tests are added. We're going to make the assumption that the secretary's advisory council has already recommended it to the rough. What's your next step?

Often times that can be done through several mechanisms in the state. You can have legislation put into place that dictates your localized test. Often times legislation has a shorter time line than sometimes you might like. Usually when it is in legislation it gets the timeliest set for you.

In my experience it's much shorter than if we did more of a grassroots approach and began the testing internally. Then asked to amend the reg on our own.

The next thing we need to do is get it into our regulation, at least in our state even if things are legislated we still have to amend our regulations and that practice is the same whether we have the legislation or not. The next thing we need to consider is your hired by your committee in your own state or whatever type of group of people make recommendations or advise your commissioners or health.

How much clout do they have? Are their recommendations usually heeded? Are they just recommendations? How does that pull those 2 the other things that have to be accomplished from a legal standpoint in order to implement a new test universally in your state?

Then the fourth thing that we often get asked for. At least we do in preparation of regulatory package is a cost analysis. One thing to keep in mind is that you might have to do this several different times for different audiences.

Your advisor committee may ask for this when they're considering if you're state should adhere to the recommendations on the rep. You may have your state commissioner may ask for such a thing to look at even before the regulation.

Keep in mind that you might have to prepare different emphasis for different stakeholders when you think about adding something to the panel. Next slide please.

The first thing that we tend to look at while we're starting on the legal end is to look at the test and look at what's known. Either methods available. Are you going to be the first one in the country adding this to your panel? Are you looking from a research type test or is there already a clinical test? Are other states doing this? If it is in research, that's the most important thing to consider is how scalable it is.

Research methods in our hands work differently when you're doing a test of only a few specimens a day. When you go to scale that up to hundreds or thousands

you might run into some more blocks. That's one of the first things that we look at is how scalable it is.

We also want to take a look at whether or not we're going to need a second tier test. In our state for example, for ALD, we run a fast HPLC test. A fast MS test sorry. Then we even have a slower HPLC MS test that's done to find the trabeculation on the analyte that's abnormal so that we're able to not refer people because there's an interference in the mass spec test the first tier that artificially elevates the results.

By running it on the HPLC with a longer retention time you end up separating that interference out. You have to take a look at that type of literature and expect it's best to have scalable tests as well. It has to fit into your work flow. You have to make sure you can handle volume and the number of referrals on the other end.

You need to also look at how much you can handle on your second tier. How many samples will go from first tier to second tier. That gets factors in many different places. It factors your work flow. It factors in your cost. It factors in the rate of referral.

Then you have to think about the future. Where are you headed in the lab with that type of test. Are you able to multiplex? Do you have a platform already in place for that? If you do do you have the capacity to add more tests on to that platform or do you have to start from scratch with a whole new platform?

When you are first thinking about how you're going to implement the test all of these things play into the methodology that you choose. In some cases you don't necessarily have a lot of choice on what method but when you do this becomes pretty important in your consideration. Next slide.

The next thing you need to look at when you're considering the types of testing that you're going to implement is your space and what your infrastructure is like. Are you going to need to construct a new room? A new building in some cases? Are you going to have to move?

In our case when we moved to a newer building we had to add backup power. You need to consider whether you need surge protectors and that sort of thing. Do you need more space? Do you need to accumulate more rooms? Do you need to get them prepared? How does that fit into your total work flow?

When I talk about work flow from a laboratory perspective it's thinking about punching. Your punching order. Your testing. How soon it needs to get started on a given day. Then you also have to consider how it flows through the laboratory. The timing of your testing. To ensure that if you do have referrals it gets out to follow up and that things can be completed in the time frame that you need for your reporting.

Sometimes this isn't a big deal. Sometimes it is a big deal. Like we saw with the addition of DNA testing for SCID many labs had to find space to be able to do the molecular testing because of the concerns about PCR and contamination and to have that kind of testing built into an important screening workflow where it might not have existed originally. For some cases this could be quite a big challenge to overcome.

Okay. Next slide please.

The next thing we consider is sometimes this comes with when you're doing your cross analysis. If you're going to get any kind of funding you do this. Whether it's grant type funding state funding or a fee increase is staffing.

You need to have expertise to do this type of test in house already. Are you going to implement a test that's included in the add on where you have people who are knowledgeable of the technology? Or are you just going to add extra burden to your already overworked staff? How can you make that happen and make it feasible in your laboratory?

You also may consider the willingness of your staff. They may need to go for training. They may have pressure due to the short timeline. Do you have a team that embellishes change? A group that will buy into the addition of a new test and really help to get it off the ground and get started?

On the follow up side you have to handle the new referrals. You have to think of staffing with respect to the entire system of newborn screening. I'll cover that in a couple of slides. You want to be concerned not only about the people who are doing the testing. You want to include people in accessioning. People in punching. People who are getting the results out the other end.

Do you have medical providers in your state? Are there enough of them? Do they have a great expertise for follow up? Are they located throughout the state that they could handle your referrals? Do you have their buy-ins? Have you engaged them early?

When I think about staffing I think about the whole system. In addition to just having the laboratory expertise to do whatever you're challenged to implement. Next slide.

When one considers funding there's several things that might have to happen. You may have to acquire new equipment. That may be making decisions whether you're going to purchase equipment. If you purchase it are you going to have a warranty? A service contract beyond the warranty?

Or does your state allow to have a reagent rental type agreement where you get the instrument because you're buying specific reagents? Sometimes those are easier. Sometimes they're more difficult.

Along with considering space and infrastructure if you need new equipment you need to figure out where you're going to place it in your laboratory.

Then personal service. Again. People who are actually hired to do that part of the task. Do that part of follow up. Or as I said earlier do you have staff that are already engaged and ready to implement the new test?

The other thing that we consider in our state is the availability of reagents. One thing that we always ask our vendors are, are you going to be able to supply our needs? We need reagents that are relatively high volume.

One of the things that we always get concerned about is how big a lot might be. Because if the lots are small and you have a full supplier you're going to spend a lot of time validating your reagents. That adds to your cost overall.

Questions for the vendor will be: How big are your lots? Are you making specific lots for my state? Are you willing to be able to keep up? What is the backup in case something goes wrong if you have a sole provider for reagents? Those can be a big problem because we can't afford to stop once we start.

We like to get that attestation from a vendor at the beginning to at least ensure that there's a system in place in the event that there's any kind of failure on the side of the vendor. Or that we have a lot that fails. How quickly that can be resupplied.

Sometimes I think people forget about that. It's an important piece to make sure that that's in line before you implement your testing.

You also have sometimes the option if you need to acquire equipment or you need to acquire reagents are they on a state contract. How much flexibility do you have if they're not on a state contract? How easy is it for you to full forest or single forest from a specific vendor? Is there a cap in place? Can you only buy X dollars reagents worth? Is pricing locked in for any period of time?

When you're thinking about funding you have several different levels of funding whether it's staff reagents and also any new equipment that you might need to complete the task. Sometimes contracts can take a really long time. It's important to start that almost at the same time you're starting on the legal end of it. To make sure that you're going to have everything in place. Next slide.

When you consider quality control and quality assurance you need to have some sort of mechanism to use to identify specimens. With the new rules regarding specimen use sometimes this can be a challenge.

It varies based on the state of the seal. It varies based on what your actual state is allowed to do. What your IRB will allow. Can you get permission to use DBS. Dried blood spots that are stored. [inaudible 00:20:48] whose children are actually effected. Then also to get your population means. Your cutoff values. Are you able to use specimens for that?

We used to be able to double punch specimens coming in the door. Be able to use that to develop a test. That has become much more difficult in light of the new changes due to section 12.

The other thing is that one is to consider early on and engage is CDC. They are really good at keeping a pulse of what's going on in the community. We need to make sure that we have ample quality assurance materials. That there's reference materials. That there's some system for proficiency testing. So that we can maintain our federal goals for laboratory testing and clinical testing.

Early on it's important to get on board with CDC and others who may be in states that may be looking at the same type of screening or who may be already screening. To make sure you have access to these types of materials. Next slide.

The other ongoing quality control that you need to have, or positive control. Here's where we've really worked well with advocacy groups and partnered with them to be able to allow parents who have children effected with some of these conditions to leave their babies samples from our depository. For our testing. To test.

We don't like to use until we've gotten pretty close to an idea of where our cutoff might be looking at the population. We found that by working with the advocacy groups and partnering with them we usually get enough parents buying in and allowing us access to their children's newborn screening specimens. They know how important they are. To have the residual specimen of a child whose actually effected and diagnosed.

We also work with our referral centers. Whether it's your SCID centers or your metabolic centers. If they have patients who have a diagnosis and they've been following them sometimes you can work through the physicians at your specialty care centers. Get permission from parents that are seeing those providers to allow use of the newborn screening samples as well.

We also sometimes are able to get samples from other states. Although this has also been hampered a bit by the new informed consent rules. Even if it's not federal funding it's sometimes a challenge to get other states who are screening to be able to share stuff and it's because of internal policies.

When you're using these types of samples and you're using them from children who are already diagnosed you'll need to implement a consent process. In our

state we get IRB approval even though there are big huge steps to identifying those babies we pass it through our IRB to make sure that we have permission to use those babies perfectly.

The important thing we're here to consider that we need to have available real controls. Sometimes a set of controls can be used. We found particularly with CRADA and some of the other conditions that it's really important to have controls from real people who have a clinical diagnosis for that condition. Get that specimen from the newborn period rather than an adult or an older child who may have that condition. The values can be quite different when you consider a newborn blood sample versus a sample from an older child.

The important thing also if you are doing any kind of DNA testing or because all these conditions are always something that's tested it's good to get carrier controls or heterozygous controls. To see how the bad chemical analyzes the hades in your assay.

Also from a DNA standpoint if you're doing that type of second tier testing to make sure that you can detect those children and get an idea of which genotypes much be common in your population. That may inform when you compare that to some of the sites that linked a phenotype to that particular genotype. You can get some information and hopefully be able to give that to providers in the end.

Positive controls are hard to come by. They are incredibly important as well. This is a challenge for programs. Okay. Next slide.

The other issue when you are setting cut offs especially for a bio chemical test is to consider all of your sub-populations. You may feel that running 1,000 samples or 5,000 samples might be enough in order to set your cut off. But you may see different populations emerge as testing goes on. Of course you may implement as conservative cut off to rule that out but as time goes on you might want to double back and reconsider how we look at sub-populations.

These can be premature births in New York. The rate of pre-maturity is about 14% so it's pretty high. Are there any variables introduced if specimens are collected early? Conversely if they're collected late?

Also the issue of low birth weight babies which may tie into pre-maturity obviously. Those low birth weight babies may have other issues as well. You might want to consider babies who are on TPN. How that might impact your biochemical results. As well as effects on antibiotics and transfusion. Again depending on which analyte you're looking at. What biochemistry you're dealing with. What part of the dry blood your actually interested in for your test.

We have examples of this from [inaudible 00:27:12]. We've seen differences in enzyme activities from babies whose specimens who were collected earlier

instead of later. We've also obviously people middle. It's been discussed on several calls.

If she's with SCID and prematurity. Where you may have a lower track count if the baby is premature. You also may have a lot track count regardless of whether the baby's premature. Meaning the baby may actually have SCID. You want to make sure you consider these different groups when you're looking in.

Even as you go through your validation if you parsed out those groups and take a look at what the means are and what the spread is under those variables that'll give you a clue as to whether you need to double back at some point and reassess what your cutoffs are. Whether you need to separate those groups out.

Okay. Number 10.

Then there's what I call all of the other last minute considerations. Which they tend to be last minute but on some of them you're possible working through those while you're doing everything else.

The things that tend to fall to the last minute that are just as important as that number one. We already talked about the regulations. That typically takes a long time. You need to check on that as it percolates through your state system. Your state health department and beyond.

Also your reporting system. How are you going to set your mnemonics. How you're going to merge your results. When you're going to merge your results. What your mailers are going to say. How you're going to report various classifications of your results. Sometimes that's not trivial.

Having that ongoing is probably as important as validating your tests to make sure. We typically operate in a test system environment for a while and try to generate all types of conditions. All types of results for a new condition to make sure that all the mnemonics are mapped and everything. All the letters are mapped and the results are mapped properly.

While this is also ongoing you're staffed from the federal CLIA act standpoint. Your staff has to be trained. They're going to have to show competency. You need to get that done. Get all of your sign offs for that as well.

The other thing is to inform all. You have to inform your hospitals. Hopefully your specialist centers have been engaged from the beginning and they know it's coming and they're informed. Your birth hospitals will need to know. The public will need to know. How long will it take for you to do that? Also, with respect to your own website. Updating other websites. We have the newSTEPS website. Baby's first test. Things like that. You want to make sure that everything is aligned.

With pushing out the new test implementation date you need to also consider whether or not you need to get approval through either your state health department or your own institutional laboratory. Director. Whatever. To make sure you have permission to post that.

Hopefully you have an idea of how long that's going to take. If you need to get that kind of clearance. How long it takes to percolate through. You don't want to announce you're starting and then not have your materials posted and available at the start date.

You do not want to be sending out results that are positive and haven't properly informed all of the stakeholders in the newborn screening system that this is happening. So it's important that all of these last minute things actually are all well timed out so that everything is ready to go on day number one. Next slide.

I'll just introduce. I touched on a lot of these with regards to follow up appointments. It's very important to engage. We found this works best for us to engage in and involve your specialty care centers. Form a task force or some group that has all the stake holders including the parents. The primary care representation. People from the laboratory. Your specialty care center director for whatever that specialty will be.

We found it's really helpful to go out and find the condition experts. Engage them early on. They're really good at helping move the process along and giving us feedback for short term and long term follow up. We learned back when we were implementing SCID and ALD that it's good to come up with some diagnostic and management guidelines and case definitions. It's really important for reporting. It helps us get everyone on the same page. Even if they get revised at some point.

The diagnostic and national guidelines that they get codes for everyone who has the algorithm and everyone knows how to approach these babies once we identify them in a laboratory. It's very helpful because it allows us to really look at the follow up. Look at the laboratory work. Go back and reassess the lab if we need to. Everyone is on the same page at the very beginning.

Follow up staff also needs full training. They're going to need to be able to communicate the results to the medical community. I understand that a lot of times the primary care providers may not know much about a particular condition due to its rarity. How they will give results out to the specialty care centers. They usually tell us how they want to receive their results. That all has to be done up front.

Then obviously the end of the day is to come up with a plan for long term follow up so we can double back over the long haul. See how good our test was. How good our laboratory results allowed us to find these babies early and diagnose and treat them. Next slide.

I want to everybody for calling again. I hope that we have some good discussion. I'm happy to answer any questions. Thank you.

Kim Piper: Hi. This is Kim Piper. I'd like to thank Dr. Caggana on behalf of the new conditions work group and everyone on the presentation here today.

I'd like to open the dock for questions. [inaudible 00:34:12] would you like to give some directions about how folks can send their questions?

Guisou: Yes. Thank you Kim. Thank you Michele for an excellent presentation.

We are happy to take questions if you have a question you may hit star 7 on your phone to unmute your line. You may also submit questions via the chat box in the webinar window on Ready Talk.

We can read your questions over the phone.

Again that's star 7 to unmute your line.

Kim Piper: This is Kim again. I'm going to start off with a question while other people get their thoughts together.

Dr. Caggana you had mentioned that it's very helpful to get the heterozygous controls. Where do you get those? Where do you get the heterozygous controls? What's a good resource?

Dr. Caggana: We generally get them from parents of effected children. When we're working with [inaudible 00:35:13]. When we were doing Krabbe in fact we went to one of the Hunter's Hope symposiums. They hold them every summer. We had some children and parents who got consent to get some blood samples. In the development also the heart testing out the assay.

Kim Piper: Okay.

Dr. Caggana: Sometimes [inaudible 00:35:35] as well. If they're older.

Kim Piper: Great. Thanks.

Do you see. Do we have any other questions submitted yet?

Guisou: I see that Kim Turner has a hand raised.

Kim if you'd like to hit star 7 on your phone or either submit your question through the chat box we're happy to address it.

[webinar participant]: Hi can you hear me?

Guisou: Yes.

webinar participant]: Hi. Yes.

This question is coming up from Virginia. Essentially how long do the ongoing FDA guidelines effect all that we spoke of earlier? In terms of using laboratory developed tests rather than FDA approved kits?

Dr. Caggana: If it's a product that you're purchasing that vendor needs to undergo FDA approval. If it's labs that at this point I'm not clear on if there's no other tests available and there's no other tests available and you're validating it in house. The FDA doesn't have the final guidance on submission of those to the FDA for approval.

I know in New York we have our own regulatory framework we need to submit our tests for validation- Our validation packages sorry. Our protocols to New York state for approval. I'm not clear on a lab developed test and where the FDA stands right now because there's no final guidance.

That's a good question. (Laughs).

webinar participant: Thank you.

Guisou: Any other questions? Please hit star 7 on your phone to unmute.

webinar participant: Star?

Guisou: Hello yes?

webinar participant: Hey this is Sean from New Jersey. How are you?

Guisou: Hi I'm good.

Dr. Caggana: Good how are you Sean?

webinar participant: Good.

With synthetic controls do you usually make them in house for LSDs?

Dr. Caggana: For synthetic controls?

webinar participant: Do you make it? Do you use any synthetic controls for LSDs? I guess not right?

Dr. Caggana: Not for LSDs. We have used depleted blood for skin. We can make some DNA controls if you need to. We've found that actually in our early testing for LSDs we found that for pompe at least that adult blood has very low enzyme activity so that will allow you to see the absence of activity. It's not easy to quantitate I guess.

We were originally doing LSDs originally starting to do our work with krabbe. We had a consent protocol that we were allowed to pull our own blood. We had our own Phlebotomist on staff. They were allowed to draw blood. We found that the enzyme levels of adults for gaucher are quite low. We all screened positive for krabbe.

webinar participant: Okay. Thank you.

Dr. Caggana: Mm-hmm (affirmative).

Kim Piper: This is Kim Piper again.

Dr. Caggana who leads the process to do reviews for the additions of new conditions in New York? Is it usually teams? Do you have a work group? Who leads that effort?

Dr. Caggana: There's usually not-

We don't have a formal advisory committee with appointed people. We tend to have our providers we work with. Obviously with advocacy for ALD and for krabbe. If we decide that there's a test that's either being discussed for the list or that there's interest in doing then we can go ahead and start looking at the test on our level.

Then typically I let my center director know. Then if she approves it we sort of start from the ground up approach. Then we'll go up to the commissioner and say, "We have a test we think it'll be useful for babies in New York."

We do it that way. I don't know that we ever had a formal work group. Years ago Kent Tab had a work group, we called it, that went over new implementation. Now with what we're looking at MCAD and CS edition so that was back in 2001 2 time-frame.

Kim Piper: Thank you.

Dr. Caggana: Mm-hmm (affirmative).

Kim Piper: Are there any other questions?

Since I'm not hearing any other questions and it doesn't seem like there's anything in the chat box I'm going to go ahead and turn it back to Julie to wrap up and talk about future direction.

Jelili: Thank you Kim. Thank you Fizza. Many thanks of Dr. Caggana for the presentation.

If you do have any questions while I'm talking here feel free to interrupt.

I do have a number of breaking news related to new conditions that I'd like to share with you all.

Pertaining to Dr. Caggana's presentation. I strongly believe that all of the things that she noted are things that most states would need to consider before implementing any one of the new conditions. After obviously they have the authority to screen for the condition.

We will be archiving this particular webinar and all of our webinars on our website in case anyone wants to come back and listen to this particular webinar in the future.

I've been instructed to know that this is part one of a two part webinar. Sorry for the fire truck going by. The second part will deal with the same kind of implementation strategies and considerations with a focus on education follow up and other relevant implementation activities related to newborn screening conditions. Look out for an email from these 2 or someone from newSTEPS related to that second part of this particular webinar series.

As most of you know APHL received a new cooperative agreement from HERSA for 2 years to be able to assist states in a number of technical assistance in supporting implementation of any one of the 3 new conditions that has been added to the RUS. MPS1 PROP and XALD.

That particular cooperative agreement started on September 1st of 2016. Approximately 41 days ago. We will as always partnering with the Carter School of Public Health as well as Babie's First Test as well as ACMG's newborn screening translational research network to implement a number of goals related to this difficult project.

No need to go into the goals but you can imagine from all of this that you just heard from Dr. Caggana once you get that authority to screen you need to figure out how to allocate resources for these new conditions. We will hopefully provide technical assistance and resources to gain authority in those states that may need help with gain authority and funding to implement any one of these 3 conditions.

Provide resources and technical assistance to increase the number of states. Now I think there are about 5 states that are screening for MPS-1 and pompe. Approximately 3 right now that are screening for XALT.

Our goal is to increase the number of states that are screening. Or achieve full implementation for at least one of these 3 conditions with the monies from HERSA as well as to assemble develop and disseminate educational materials for these relevant new conditions in newborn screening.

As of today October 11th there will be 2 requests for proposals that will be at least 2 state newborn screening programs across the country. As you all consider adding any one of these 3 conditions to your own recommended panels.

The first we are seeking applicants to implement one of the 3 conditions. Programs can apply for a sum of funds from us on a yearly basis for the next 2 years. Right now we are looking at approximately 70 thousand dollars.

That rsc as I noted might not or will go out today that will be a blast email going out to everyone. Being that it a request for proposals then we have to do this in a competitive way it will be on our public facing website. If you actually go right now to aphl dot org slash r f p you will be able to see the active links with more pertinent information as it is related to the rfp.

I described the first one. Rfp number 2 we will be seeking applicants to serve as peer network resource centers to provide technical assistance and or bursts and or second tier newborn screening for any one of those 3 conditions.

Programs can apply for up to at this point there will be a limited number of these peer network resource centers. 60 thousand dollars to be able to provide the noted technical assistance first or second tier screening for pompe and NTS1 and XALT. That rfp also is on aphl dot org slash r f p.

I will also be on our newSTEPS website and we will send a blast email. You will receive a blast email from Sika saying our manager on newSTEPS sometime at the 3 o'clock eastern hour with more information about this.

There will be a technical assistance call related to this particular initiative. Correct me if I'm mistaken Sikha I think it's October 18th correct?

Sikha: Yes. The technical assistance webinar is October 18th.

Jelili: Okay. For anyone who has any questions related to anything or any one of these rfps we will have a webinar to talk about objectives. What we plan to gain from it. I told you some of our objectives and our goals.

We're trying to expand a number of states that are screening. I think we are in a very interesting time here where this I think is the largest. Or will be over the next couple of years. The largest expansion of state newborn screening panels since the HENG HERSA recommended uniform screening panel. The first 29 conditions that we have.

These 3 new conditions plus another 7 states that are still finding ways to implement severe combined immunodeficiencies. Listening to all the things Dr. Caggana spoke about. All the strategies that are going to be needed in place once you have the authority to screen. We want to be able to provide all those resources and technical assistance through these fundings from HERSA to you all at the resource center for state newborn screening programs.

Do we have any questions? Specifically related to the rfp? The lead is Sika Singh or you can just email newSTEPS at APHL dot org for any questions.

I'll pause there if anyone has any questions related to Dr. Caggana's presentation.

Speaker 8: Question from [inaudible 00:49:53] for Dr. Caggana.

This is Richard Haughton Again. Just wanted to know what current PT materials are available through NSQAP as well as whether second tier materials are currently under development?

Dr. Caggana: Somebody CDC is on the line. We do get proficiency materials from CDC for all of the conditions that currently are on the rus.

Speaker 8: Okay.

Dr. Caggana: As far as CNA they're obviously with cystic fibrosis they're working on some of the other conditions as we speak. Those are coming.

Speaker 8: Okay.

Dr. Caggana: I don't. We do internal Pcs for krabbe. DNA that is. We currently do internal ALD.

Speaker 8: So if a lab were to launch one of these assays or screening methodologies use them molecular second tier if CDC doesn't have the proficiency materials already made is it possible to have some intra-laboratory exchange of materials then to assess blinds to samples?

Dr. Caggana: Yeah we've been able to share with some programs some controls. Some control punches from individuals. From our CLIA teaching programs we do internal blinded teaching.

Speaker 8: Okay.

Dr. Caggana: Both of those are available.

Speaker 8: Thank you Michelle.

Dr. Caggana: Mm-hmm (affirmative).

Jelili: Any other questions for Dr. Caggana? Please press star 7 to unmute yourself or type in in the text box.

If there are no questions ... Again many thanks to the new conditions work group. All of the members of that particular work group. The co chairs of the work group. The new [inaudible 00:52:09] committee. Dr. Caggana for her presentation as noted this is one of 2 part webinar that we will have on the strategies and the next one will focus and follow up on education activities for implementation of new conditions.

We look forward to engage in the community in getting more ideas eon what kinds o activities and discussions that you wold like us to bring forth to the newborn screening community so that we can discuss. If there are no other questions thank you and good afternoon.

Guisou: Thank you everyone.

Dr. Caggana: Thank you.