Newborn Screening for Critical Congenital Heart Disease: Current Implementation Status and Future

Dr. Russell Kirby:

Who my colleague will introduce, but I just wanted to say a few words of welcome. This webinar is a collaboration between the NewSTEPs program at the Association of Public Health Laboratories, the National Birth Defects Prevention Network and colleagues at CDC, and the National Center for Birth Defects and Developmental Disabilities. And we're hoping that you'll enjoy the program and learn some things and hopefully ask some questions or get some answers. And we do plan to have actually a series of webinars. This is our first. So we'll tell you more about what our plans are as we go along. And I'm going to turn things over, to do more formal introductions, to Jill Glidewell from CDC.

Jill Glidewell:

Thank you, Dr. Kirby. Hi everyone, I'm Jill Glidewell. I'm a health scientist at CDC's National Center on Birth Defects and Developmental Disabilities. And I'd like to echo Dr. Kirby's welcome to all of you. At the end of today's webinar, participants will be able to describe the implementation status of newborn screening for CCHD in the United States, describe primary and secondary targets and locations for public health programs and to identify data infrastructure, linkages, and collection and educational opportunities with regards to CCHD newborn screening. Today's presentation will be recorded and all participants will be muted during the presentation. And again, as Russell mentioned, we'll have three speakers that I will introduce prior to each of their sections of the presentation. And we'll hold all questions till the end. But if you do have a question along the way, please feel free to type it in the chat box.

And with that, I will introduce today's first speaker Careema Yusuf is a manager for the newborn screening technical assistance and evaluation program or NewSTEPs at APHL where she manages the overall activities related to the website and data repository system for NewSTEPs. Careema provides technical assistance and support related to quality practices, data analysis, and reporting. She serves as the staff liaison for the NewSTEPs CCHD data response team. She has worked in a variety of public health settings that have included working at the state of Maryland on HIV/AIDS prevention program performance measures, and with Johnson Bassin and Shaw international working group on quality improvement activities with substance use treatment programs in the US. Careema holds a master of public health from the George Washington University in Washington DC. And with that Careema, I will turn it over to you.

Careema Yusuf:

Thank you so much. So good afternoon, everyone. And welcome. I am delighted to be here with my co-presenters, Lisa and Amy, to present on the current newborn screening implementation status of critical congenital heart disease and future needs. We'll begin with the importance of CCHD newborn screening, and then we'll delve into the landscape of screening in the US what is happening at the national level, some of the data and resources that are available through NewSTEPs. We'll also describe newborn screening programs and their activities around screening for CCHD. How is it implemented, the evolution of screening targets, what is considered a newborn screening detection of CCHD, and the different screening algorithms. We will end today with future needs, including those around data infrastructure, collection, education of family, primary care physicians, and also provide you a brief introduction into clinical considerations of CCHD, which is a primer to the next webinar. In this series.

I'd like to begin by sharing the story of Veronica Easley. Veronica is the daughter of Olivia Easley, a physician and mother who has been an amazing advocate for CCHD screening in the state of Maryland and throughout the US. Veronica was thought to be a healthy newborn and did not show any signs or symptoms of her critical CCHD before she passed away in her sleep at seven weeks of age. So of the infants on the screen, which would you assess to be cyanotic? It is very hard to detect cyanosis, which is a symptom of CCHD by just looking at the infant. Pulse oximetry helps us to find those infants with cyanosis earlier and allows for quick and timely diagnosis and treatment. The baby on the left is Veronica. The baby on the right is the first baby with CCHD to be detected by newborn screening at the Holy Cross Hospital in Maryland, just before he was discharged because of his newborn screen, he was able to get diagnosed and treatment in a timely manner. And just this last week, his mom shared that he's doing very well and is now in the third grade.

So what is newborn screening for CCHD? So pulse oximetry allows us to screen for cyanosis using red and infrared light to measure oxygen saturation of blood. Simply taping a light source and center on the extremity allows a noninvasive and painless way to measure oxygen levels in babies. This method detects for other conditions associated with low oxygen levels, including critical congenital heart disease. Newborn screening programs are state-based public health programs and screen for conditions on their state advisory committees, as they deem appropriate. At the national level, there is an advisory committee for heritable conditions in newborns and children that recommends conditions to be added to a recommended uniform screening panel or RUSP.

The recommendation process includes an evidence review, a vote by the committee to add the condition to the RUSP, and the review and acceptance of the recommendation by the Secretary of Health and Human Services. Once the condition is on the RUSP, each state or territory makes a decision to add the condition to theit screening panel. So for critical congenital heart disease, screening was endorsed by the Secretary of Health and Human Services in September, 2011, The American Heart Association, The American College of Cardiology, The American Academy of Pediatrics and The March of Dimes all endorsed newborn screening for CCHD.

So as of today, 53 newborn screening programs are university screening for CCHD. A study published in The Journal of American Medical Association in 2017 showed that the adoption of mandatory screening policy was associated with statistically significant decline in mortality of about 33% due to this CCHD mandate in place compared to those states without such policies. So again, just showing you the importance of having the CCHD screening policy. There are unique challenges and opportunities with CCHD newborn screening implementation. A few of these are listed here, for example, issues around data collection, linking with partners like birth defects registries, and then the education piece. Throughout the presentation, we will discuss these in more detail. As a national newborn screening resource center, NewSTEPs provides data and technical assistance and training to newborn screening programs and assist them with quality improvement initiatives. The NewSTEPs website has several resources focused on CCHD newborn screening, including archived webinars, educational tools, and all of these are freely available for your use.

When screening programs do submit confirmed cases for critical congenital heart disease, identified through newborn screening into the NewSTEPs data repository to date, there are 12 programs contributing such data, and this shows the breakdown of the types of CCHD that they are reporting and more on the different newborn screening targets will follow later in this presentation. So NewSTEPs also provides technical assistance with the NewSTEPs CCHD technical assistance work group, and this work group was originally created to support the newborn screening community in their efforts to implement CCHD screening. And as of July, 2018, all programs have requirements in place to screen for babies for CCHD. So, the work group priorities have shifted now to assist states with data collection and data analysis. And this group functions now as a CCHD data response team.

The group provides technical assistance around data collection, data definitions, asking questions around what data is needed, determining the line between the need to have a program perspective and the nice to have from an analysis of clinical perspective. The group collaborates with stakeholders like birth defects registries, and on quality improvement efforts. The team includes representatives from programs, advocacy groups, clinical partners, and our federal partners at the Health Resources and Services Administration, as well as The Center for Disease Control and Prevention. And now I'd like to turn it over to Lisa to provide more details on the actual implementation of newborn screening for CCHD.

Jill Glidewell:

Thank you, Careema. Lisa Hom is a nurse at Children's National Hospital and has worked for the past 10 years in the heart Institute. She's also co-chair of the NewSTEPs data response team. Prior to starting her work at Children's National, she had the unique opportunity to work both as a regulatory counsel, focusing on healthcare issues at the state level and as a pediatric intensive care nurse working directly with children with CCHD. Lisa lives outside Washington DC, and holds a Juris doctorate as well as Baccalaureate degrees in nursing and history from George Mason University and from The College of William and Mary. Lisa?

Lisa Hom:

Thanks so much, Jill. And thanks Careema for that really wonderful overview of implementation and education at the national level. And certainly we've accomplished a lot since the US map looked like this beginning in 2018 with every state having a CCHD screening program implemented. However, when it comes to critical congenital heart disease screening, if a nice map like this or a picture is worth a thousand words, what we also find with screening is that the Devil is often in the details. And I actually stole this from my co-chair Amy. I really think that this really captures well, what we see when we really take a closer look at how states have implemented critical congenital heart disease screening. And when we do that, what we find is that for these five reasons, critical congenital heart disease screening using pulse oximetry is actually one of the least uniform conditions on the recommended uniform screening panel.

And it might actually look a little bit more like this patchwork quilt. If we really look at the state variation and implementation, and it's mainly for these five reasons, first they utilize various definitions for primary and secondary targets. Although most states do capture a core group of primary targets, states vary in their authority to collect data and in their ability to analyze that data. They vary in their integration methods with other programs such as birth defects programs, as well as the population screened and in the implementation of which algorithm they use in their hospitals. So, newborn screening is unique to all the newborn screening conditions on the RUSP and unlike the others, I don't think any others have two other lines of defense or other mechanisms during the newborn period, which can detect or identify infants who may have disease. So also in addition to pulse oximetry screening, it's sort of the third line of defense.

There's the prenatal ultrasound, as well as the clinical assessment that occurs in that first few hours in the first day of life. Certainly other public health conditions are identified and in most states the primary critical congenital heart disease targets are being reported either through newborn screening or birth defects registries. In addition to the impact of the screen itself, definitely can vary both by individual and by location. What we see when we take a look at the national landscape is that certain parts of the country have much higher rates of prenatal detection, as well as what clinical care is available and accessible to an infant based on where it's born.

So what can we agree about nationally? And certainly there is a sort of uniform definition of what a critical congenital heart defect is nationally. We agree that it's a group of serious heart defects in this critical form typically have low blood oxygen levels in the newborn period they're conditions that require intervention either soon after birth, some definitions say in the first month of life and some say in the first year of life, and those interventions are typically cardiovascular surgery or cardiac catheterization, and they may or may not be ductal dependent. And why does that matter? What does ductal dependent mean? So this is just an illustration of one of the most deadly and one of the most common forms of critical heart defect and that's Hypoplastic Left Heart Syndrome. So, in that first 24 hours age on the photo on the left is a picture of what sort of normal newborn circulation should look like after the infant has transitioned from fetal to newborn circulation.

And what we see is sort of nice blue and red blood kind of enclosed circuits circulating the blue blood is going out to the lungs. The red blood is coming back, it's oxygenated, and it's going out to the rest of the body. What we see in infants who have a serious heart defect is that there is a mixing of the blue and red blood that presents as cyanosis or low blood oxygen levels. And that's important because we try to screen infants at or around 24 hours of age, where we can see that mixing of the oxygenated and deoxygenated blood. And so this infant would have a failing pulse oximetry saturation of 90% or less in that neonatal period.

So Careema touched on what pulse oximetry is as a screening method. Certainly it's painless and noninvasive. And this is just a picture of an infant being screened by pulse oximeter. What we know about pulse oximetry screening is that it has moderate sensitivity, and this is based on a meta analysis published out of England in 2018, where they examined all different algorithms and found good sensitivity, specificity, and a low false positive rate. So in 2011, when critical congenital heart disease screening with added to the RUSP, and there were seven original targets published, and it's this list right here. And those were the primary targets that pediatric cardiologists felt were the ones that were most likely to be detected by screening after which there were other lists and one of those expanded list included these additional five lesions, interrupted aortic arch, coarctation, Ebstein's, double outlet, right ventricle and single ventricle. And these were an additional five lesions that can present with cyanosis and be detected as a result of critical congenital heart disease screening. Some lists also include aortic stenosis and pulmonary stenosis. So I have those up here as well.

So this list right here was published in 2016 and Jill was one of the authors and part of the expert panel that was put together. It was a joint panel with the American Academy of Pediatrics and they published this revised list. And this list important because it includes the original seven primary targets, as well as the five expanded targets. And then they also included. And if you can see the fine font here, but other critical cyanotic lesions, not otherwise specified, and that can be the AAS or the PS as well as other cyanotic heart defects. And what makes it difficult is that if we don't sort of have a common uniform agreement in terms of what those primary conditions are, it can really impact our ability to analyze outcomes and really get a good grip in terms of what the sensitivity is and what that newborn screen program can say about their outcomes for heart defects detected by pulse oximetry screening.

Also importantly, this list included the secondary conditions and these are other reasons why an infant might have a low blood oxygen saturation level in that neonatal period. And some states do track and count these secondary targets for screening as findings. Certainly they're not a true positive for critical congenital heart disease, but they're very important to detect. And some in most cases they're detected more frequently than critical congenital heart defect. For every one case of CCHD, public health programs are often finding three or four cases of these, what we're classifying as secondary targets. So these are infants that are failing for low blood oxygenation stats for other reasons, including pneumonia, sepsis, and perhaps a non-critical form of congenital heart disease. When we look globally

around the world, what we find is secondary targets and where we're sometimes able to make the most headway in terms of lives saved. And that's particularly true in parts of the world where infants might not have access to cardiovascular surgery.

So in addition to what programs are tracking and calling primary and secondary targets, we also see a lot of variation in terms of what data is being collected associated with pulse oximetry screening. What's sort of nice to see is that the majority, sorry, went a little too fast. The majority of programs are collecting individual level data. However, even within that group, it's kind of a narrow majority. We're seeing some variation in terms of what they're actually collecting, whether it's just a simple pass, fail, or not screened, whether it's just the final oxygen saturation levels or whether it's all of the data.

And then when we look at states that are collecting just aggregate data, I think this quote, I included it because I think it gives a good sense of how it can make it really difficult for folks in public health to get a good grip on what is happening with those abnormal screens. So this is from an email someone sent to me, where they talk about how they get no data on what actually happens with abnormal screens and that they often know when there's a false negative for CCHD screening. So when an infant that was not identified prior to discharge, and this person said that they sit on the death review committee. So they know when they, they have false negatives when infants are not identified. So they know when it doesn't go well, but it would also be nice to sort of have outcomes and follow up on times that it does go well when that safety net does work and infants are identified through screening.

This graph is just to sort of illustrate how difficult it can be. Even for states that do collect individual level data. This data's a little bit outdated, but it comes from California and California is one state where individual level data is collected. However, the public health departments, as there's a lot of variation in terms of which hospitals are reporting and that because it's an unfunded mandate, what they're seeing on any given year is that right around 60% of hospitals are reporting on births in terms of whether the screening, in terms of the individual screening data.

There's also variation amongst programs in terms of what is being counted as a screening find. Some states in their laws require all infants to be screened. So in some cases how they capture those infants, who are prenatally detected can vary. In addition, whether or not infants who had an echo and so the echocardiogram is really the gold standard in terms of being able to make a diagnosis in why the infant is having a low blood oxygen saturation, whether in terms of whether the infant. Low blood oxygen saturation, in terms of whether the infant truly has a critical form of congenital heart disease.

And then there's also instances that we see in terms of an infant, having some signs and symptoms of potential critical congenital heart disease, and then they might do an early pulse oximetry screen. And so if they do, should that be counted as a clinical assessment find, or should it be counted programmatically as a screening find? And so there can be some gray areas, in terms of how these finds or targets are categorized, and the determination of how states are answering these questions can certainly impact the analysis.

I do want to switch over now to integration and the variation, in terms of integration with state birth defects surveillance. So this data is coming to us from a NewSTEPs survey that was done several years ago, and Amy will touch on this a little bit more in our presentation. But what it found as an overview is that 19 states did have some integration and that the majority, or 32 did not. And sorry, that includes DC, that's how we get to 51. And so within those states that don't have any integration, the reasons for that vary somewhat. And so for some states, it was because no individual screening data was available to be able to link. In some states, there's no birth defects registry or surveillance. And then for 15, the systems exist, but were not linked. And I think that's where we can see some real opportunity.

Now, for those states that do have integration, what does the bi-directional communication look like between newborn screening programs and birth defects programs? And so there can be a lot of variation. What we found is that it can look like anything from kind of phone-a-friend to sending an email or a report to some states that have sort of integrated health information system.

And also for those states that are able to integrate, what are they doing and how are these data being used? And what we found is that it primarily falls into three main categories that programs are using integration to identify false negatives from screening. So those are infants that were not identified through screening because sensitivity is only in the seventies. We do certainly expect that not every infant who has critical congenital heart disease will be identified through newborn screening. It can also be used to match and identify cases as well as to help in the follow-up of failed screens and determination of what the final diagnosis or outcome was for that infant.

So what we found in seven superstar state programs, they're able to use, we have the ability use the integration for all three of these potential uses, which is very helpful. And then other programs, we see they're able to do some of these things with their integrated data set in terms of identifying missed cases, following up on outcomes or longer-term follow-up and then identifying and matching cases.

Sorry, going a little bit fast on that one. There were two state reports that did allow for comparison with birth defect registries. And these are just two examples. And in state A, there were 22 infants identified with CCHD through newborn screening, however, 230 were reported through critical congenital heart disease. I'm sorry, 230 were reported through birth defects surveillance with CCHD. And in state B, there were eight identified through newborn screening and then 561 reported through birth defects surveillance.

And so one thing that we're hoping to discuss is whether and how infants are identified, whether there's a mechanism to capture the timing in terms of when an infant was identified, whether it was in the prenatal, perinatal or postnatal period in order to help with linking the newborn screening finds with what we're seeing in birth defect surveillance.

So other sources of state variation that I touched on earlier is the population that's being screened in each state. So what we can definitely agree on is that critical congenital heart disease screening is meant to capture asymptomatic infants in a well baby nursery, prior to going home from the hospital.

And the variation that we see is when we look at individual state implementation, in terms of how states have defined this eligible population. And some states, in their law, they simply say that all newborns are required to be screened. And in other states, we see exemptions or carve-outs and some states they call it "physician discretion," in terms of whether we are screening NICU births, infants in the ICU, births that occur at altitude, or home births. And I'll just touch very briefly on these three sort of main categories.

So what we've learned in terms of screening infants in the neonatal intensive care units, we've learned, certainly, that pre- and post-ductal oxygen saturations are similar to saturations in late and preterm infants. And so a lot of the NICU protocols will require screening after the infant is around 35 weeks gestational age, and certainly can be safely implemented into a NICU.

Roughly a third of the infants in the ICU receive echocardiograms. Many of them are on oxygen and many of them are on pulse oximetry. However, what is not happening typically or prior to implementing CCHD screening is moving the probe from the right hand to the foot and getting a read on what the differential is, whether there's a difference of three or more between the hand and foot.

Two states in particular, New Jersey and Michigan, have studied screening in the NICU more closely. And what we've found from both of those published and unpublished studies is that it certainly

is to lower yields. And so I think in New Jersey, they've screened over 300,000 infants and identified one case of coarctation through newborn screening. However, that's what we would expect to find. Certainly, these infants are receiving a higher level of care. And so some states do require that these infants still be screened based on the legislation and the law, but these aren't really the infants that we're targeting.

Other states have exemptions, such as Colorado, for screening at high or moderate altitudes. And what we see is a much higher false-positive rate once we get to above about 6,000 feet. And for that reason, Colorado passed a law requiring all hospitals to screen below 7,000 feet using pulse oximetry. And that's for these four reasons listed right here.

So what did Wisconsin and the Netherlands have in common? So, the picture... Certainly one thing they have in common is that they're both well-known for cheese. However, another thing that they have in common is that both have developed screening protocols for out-of-hospital births. And what that usually looks like is an initial screen will be done before the midwife leaves the home in the first hour or two of life. And then they'll get a second screen about 24 hours later. So they'll either come into a pediatrician's office or the midwife will come back at about 24 hours of age to do a second screen. So states do vary in terms of implementation and how many home births they're seeing within their state.

So, in addition to a unique home birth screening protocol, and certainly there are several NICU protocols as well that have been published, there are a lot of algorithms nationally and internationally that are well-studied and that have variations in terms of timing and in terms of cutoffs for pulse oximetry saturations. Then which one is the best?

Well, there's a lot of data. And the three that we see that have been primarily studied and implemented in the US are what is called, the first one that's most predominant, is the American Academy of Pediatrics' Recommended Protocol. That's sometimes also called the Kemper Protocol. The vast majority of states do use this protocol. There are two others, however: the New Jersey protocol and the Tennessee protocol, I think Minnesota follows the New Jersey protocol, that are also being used in the United States.

This is a quick map that NewSTEPs put together, along with the American College of Cardiology, just to illustrate there are 10 states and those are highlighted in purple that are required, it's actually written into their law, that they're required to follow the AAP protocol. An additional 16 states either recommend or reference in some way, the American Academy of Pediatrics' Protocol. And then the remaining 24 states don't specify a protocol in state law. However, many of them have implemented the same protocol. And then I included a link. If you're interested in more detail about this, there's a letter to the editor that you can read associated with the AAP recommended protocol and US implementation.

So in an effort to incorporate some of what we've learned from screening for about 10 years in the US, a group came together and published this recommended updated strategies for pulse oximetry screening in the United States. And they took a look at published state reports, some of the data in the NewSTEPs repository, and other population studies published globally to see if there are any things that we could recommend to change in the US screening protocol or the AAP protocol that was published in 2011.

So it does capture some of the things that we've learned and sort of discusses it, however, it has not formally been endorsed. And we will continue to examine and integrate what we've learned from these 10 years of screening and what published data we have.

Want to say... This is my last slide, and I just want to say that certainly there are many successes with critical congenital heart disease screening, and it is offered in all 50 states. Nationally, we are

seeing fewer US deaths. I believe Careema mentioned a 33% reduction in states that require critical congenital heart disease screening. And that was in an early outcomes paper. I think it looked at through maybe 2016. However, what we definitely know is that outcomes of screening are still very hard to quantify, and there are differences in how state programs define their primary and secondary targets, their eligible populations. There are differences in how they collect their data, and certainly in which algorithm they've implemented.

That's everything I had. Thank you very, very much for this opportunity. And with that, I will turn it over to my co-chair Amy Gaviglio.

Jill Glidewell:

Thank you. Lisa. Amy Gaviglio is a certified genetic counselor and public health genetics consultant who has been working in the newborn screening arena for the last 13 years. She's currently a consultant with CDC, APHL and Expecting Health. She's also co-chair of APHL's new disorders work group and CCHD data response team. Amy is a member of several of APHL's work groups, including short-term follow-up, legal and legislative affairs, and health information technology. She also serves as chair of the newborn screening expert panel for the Clinical and Laboratory Standards Institute, chair of the executive subcommittee for Minnesota's Rare Disease Advisory Council, and is on the advisory committee on heritable disorders in newborn and children's education and training work group. And Amy, we will turn it back over to you.

Amy Gaviglio:

Excellent. Thank you so much, Jill. And thank you Careema, and thank you, Lisa, for setting the tone for the final part of the session, which is to focus on how we can continue to grow and evolve pulse oximetry screening across the nation.

And this part is really, it's not meant to dismiss the successes thus far. It is absolutely a success that all states have some legislation around pulse oximetry screening. It is a success that the enacting of this legislation, even in and of itself, has reduced morbidity and mortality. And certainly, it is a success that this screening program has saved the lives of newborns that would have otherwise been undetected.

But I'm going to lean on the wisdom of Lily Tomlin, who once said that the road to success is always under construction. And so when we think about the future needs for CCHD screening programs, we can really categorize them into these three primary areas.

So the first area is one where I will spend the most amount of time today. And that is the topic of data collection, integration, and analysis. As Careema and Lisa have already discussed, the variability amongst programs has really led to a lack of coordinated data collection.

And the second area is one that quite frankly, truly spans all screening programs, and this is the ongoing need to provide individual follow-up services and provide training and education to all relevant stakeholders.

And the final topic area, which I will only touch on briefly, as the next webinar in this series will cover this in more detail, is the intersection of other detection modalities, potential other biomarkers, and overarching clinical considerations. So let's start with looking at CCHD screening, data collection, integration and analysis.

So as I thought about the best way to convey the issue and need, I kind of finally settled on using the peer-reviewed literature to tell a bit of a story. So on this slide, I used Scopus to illustrate the number of publications directly related to pulse oximetry, newborn screening for detection of CCHDs.

We started in 2011 when CCHD was added to the RUSP and we can see, not surprisingly, a sharp increase in the first two years after this occurrence and then followed by a relative plateau across the next several years.

Of course, we still have a few months left in 2021, though I think it probably won't be too much of a surprise if we do see a slight dip this year in publications around this program, just due to the ongoing pandemic. But let's actually delve deeper into this data.

So if I filter the publications to only show those with domestic or US affiliations, the number of publications drops by over 50% from 268 to 92. And I think perhaps most interestingly, we really have to kind of go down to the seventh and ninth positions before we start to see public health program affiliation. So we see here, New Jersey Department of health and Wisconsin School of Medicine and Public Health with five publications each. And so, why is this? Why has there been such a lack of data coming from public health? And more importantly, what do we need to do to change this trend?

I know Lisa has already discussed some of the issues, but let's kind of condense it down further so that we can better, I think, appreciate the reasoning behind the needs that we'll present in a bit. So one ongoing reason that we see from programs in terms of understanding this program is some general lack of clarity on the responsibility of public health and the purpose of data collection at the public health level.

So there are typically four or so overarching possible reasons for data collection. And of course, depending on state statute and often resources available, a program may choose to collect screening data for any one or combination of these reasons. So programs can choose to simply use this program as another method of surveillance. So in these cases, programs may only collect data on failed screens in order to get an idea of how failure rate as well as an idea of birth prevalence of various CCHDs in their state. Or, programs could decide that they want to more robustly assess screening performance, either on the backend, so maybe getting reports on a weekly or monthly basis from birth facilities, or they could do this in more real time by either attaining pulse-ox data on the newborn screening blood spot kit card, or via some kind of electronic messaging method.

And finally, some programs may even choose to expand beyond this and collect some ancillary data, things like maybe perfusion index, or maybe the results from all the screens and not just the final screen in order to offer some contributing data to either programmatic or algorithmic improvement. But when the reason, or the "why" isn't clear of why we're collecting data, the "what", the "what" data elements need to be collected. It can't really be fully fleshed out. And I think this concept will play into some potential solutions that we'll discuss in a bit.

So the second contributing factor to a lack of data lies in roles. Where some state public health programs have certainly taken on the role of overseeing collection of data, others have not. In these cases, where does the responsibility lie? With the birth facilities themselves? Other public health programs like birth defect registries? Do the tertiary cardiology care centers take it on? Someone else? A combination?

I think programs, and what we've seen in our work over the years, kind of providing technical assistance to programs, is that those programs that have been most successful in program implementation and data collection have clearly outlined roles and responsibilities. And the, I think, important point here is that there isn't one right way. Programs can choose, I think, what way works best for their situation, their restrictions, their resources, as long as there is clear understanding and buy-in amongst all stakeholders.

So the first two factors focused on stakeholder understanding roles, responsibilities. Well, this factor surrounds the significant data silos that we see in public health. So Lisa mentioned, I think she had

a version of this graph in her talk, that over half of CCHD programs are not linked to a birth defect registry. And certainly acknowledging that not all states have birth defects registry, these numbers, it's still a tremendously missed opportunity for obtaining a better understanding of CCHD in the US.

In addition to data linkages between pulse-ox or CCHD screening and birth defects programs, linkages between vital records, both looking at birth certificates and death certificates are invaluable to data collection and analysis as it pertains to CCHDs. And this is really both from the ability to know the appropriate denominator. So who are your eligible infants? How many of them exist? But to also understand screening rates, as well as mortality from CCHD.

We did want to note, you can see those in the bottom left-hand corner, that there is a webinar scheduled on September 14th addressing vital records linkages and newborn screening. I'm hoping either Careema or Sari can put the registration link for that webinar in this chat, but we do hope you can join for that as well. Really just talking and hearing from programs who have linked to their vital records and how they've done that and the value of doing so.

So the final factor that I'll mention today, again, and why we don't seem to see a lot of data coming out of programs, is I think an overarching sense of recognition in funding towards CCHD screening or kind of lack thereof as compared to other screening programs. So EHDI, early hearing detection and intervention, and the dried blood spot program.

Lisa in her talk used I think the two words we all hate to hear in public health, which is unfunded mandate, and this has been unfortunately a true situation for many CCHD programs. So unlike the EHDI programs, they have their CDC and/or HRSA annual grants. They receive those routine grant money. CCHD implementation and improvement has really had to rely on ad hoc grants for specific work. Likewise, if you look at the dried blood spot screening program, this program is typically funded through the kit fee or the sale of the screening cards, and with only some states having incorporated paying for CCHD programmatic activities using this mechanism. And I think this lack of directed funding has led to CCHD often being done a bit as an aside, I would say, to other programs with no really dedicated national meeting to these efforts either which probably ties back to that funding issue as well.

So given this background on the why, how can we begin to improve CCHD screening data collection at the national level? So I'm going to start with the issue of funding. I mentioned that ad hoc funding opportunities for CCHD have been really important for work around this, and we do want to mention that CDC is funding a new project to collect state data on timing and mode of critical CHD detection. So this project is funded as a component of a larger notice of funding opportunity on advancing population-based surveillance and birth defects, and funding began in 2021.

So the goal of the critical CHD component of this NOFO is to understand the timing and method of CCHD detection, the disparities in timing of detection, and whether timing of detection is associated with newborn health care utilization. So funded sites, as you can see CDC has funded the eight health departments shown here. We'll conduct surveillance on critical CHD cases. It will ascertain timing and method of critical CHD detection, such as prenatal detection, clinical detection using signs and symptoms at birth or detection through pulse oximetry, or detection after discharge. They will also ascertain individual level CCHD screening results and timing of confirmatory echocardiogram. So again, we've listed the eight health departments who received this funding earlier this year, and we're certainly looking forward to hearing more about their progress in the future.

So the one thing we also really need to enhance is our focus on standardized and minimal data elements that each program should aim to collect. So in 2013, which is eight years ago, it's just amazing to think. A minimal data set was published by Dr. Martin et al, but this really never seemed to take hold. It is certainly, I think, time for us to take our experience thus far, our lessons learned and re-look at this minimal dataset. We need to enhance this work by providing better guidance to programs on not only

what data elements they need to collect, but harmonize and standardized how this is being collected so that we can actually facilitate some aggregation of this data at a regional or national level. And then I think maybe equally as important is taking this work a step further, we really need to work on dissemination and promotion of these data elements so that each program ideally in collaboration with their birth defects registry understands the data elements needed to assess the effectiveness and success of pulse oximetry screening.

Certainly a good starting point for defining a minimal data set is to start with determining what questions we as a public health programs should be able to answer around pulse oximetry screening. So we've listed a few here to get you all thinking. Clearly we should be able to determine the screen rate, what percentage of eligible newborns are getting screened? We really need to understand the failure rate. What percentage of newborns are ultimately failing the screen, according to the utilized algorithm and requiring further workup? We should understand what we are picking up with this screen, what defects are we finding? But we should also understand what we are not detecting by the screen and why. Is a case not detected due simply just to the nature of the defect as I think Lisa very nicely pointed out, or is it due to a failure of the algorithm?

How are CCHDs being detected in each state? You've heard a little bit about detection modality, and the idea that pulse oximetry serves as kind of a third line fail safe. So really understanding how this screen fits into the other detection modalities helps to understand the CCHD detection environment really more holistically. Certainly likely there are others as well, and the CCHD data response team is looking to put together a small work group to really focus on this further, and we certainly invite any additional thoughts on these questions on minimal data elements.

So to be able to assess detection rates in these cases, we certainly need more granular public health case definitions, as well as a harmonized understanding of the sensitivity and specificity of pulse oximetry screening for each distinct defect. This slide is very busy, I know, it's not meant for you to look at everything, but it's mostly meant to impress upon you I think the complexity of these definitions and how even a single defect, say Tetralogy of Fallot may fall into different categories depending upon severity or whether there are accompanying defects.

These cases themselves will also likely need to be sub-categorized in order to fully flush out and understand the screening process. So you can see here that there exists at least four potential categories for a miss case dependent, again, upon the severity of the defect, the presence of co-morbidities as well as adherence to the screening algorithm. So with some defects, if the algorithm is followed correctly, the case may be deemed a physiological miss case simply because the child was not hypoxic at the time of the screen.

In other cases, it's illustrated in the third row, the screen may simply be unable to detect the defect because it does not present with hypoxemia in the neonatal period and severity is more mild. In these cases, it is helpful to document the outcome, but maybe not consider it a missed case. Of course, in any instances where the algorithm was not followed correctly, this certainly presents an opportunity for training education, potentially process changes, which we'll touch on briefly in my next section.

So without a doubt, creating inter-agency data linkages remains one of the key areas where we need to see movement. The potential that is created, especially with linkages between CCHD programs and birth defects is as I mentioned, tremendous. In this graph, which you saw in Lisa's slide as well really shows the potential. Collaboration between the programs allows really for three kind of significant assessments. It can help screening programs obtain the outcome after a failed screen while coordinating follow-up and reducing burden on the family and providers. It can help screening programs identify affected newborns who pass the pulse oximetry screen, so so-called missed cases or false negatives. And

finally it can serve as a data quality check between programs to ensure that all affected newborns are appropriately captured by each program.

So before moving on, I just want to quickly show on one slide the summary of some needs around data collection and analysis as I know we've touched on quite a bit of information already. So standardized data recommendations, improve case understanding and definitions and enhanced inter-agency data linkages are all areas where we can put our effort to greatly improve our understanding and collection around pulse oximetry data.

So in the previous section, obviously we focused a lot on what data to get and maybe how to get it. For this section, we will talk a bit about what to do with the data outside of data analysis. And so we'll also touch a bit on broader educational and training needs for relevant stakeholders as well. So an ongoing question we in the CCHD data response team get is what should we be doing with CCHD screening in terms of follow-up? And this question arises largely again, because of that unique nature of this screen as compared to dried blood spot and EHDI.

In those programs, we are used to really pushing or dictating the process. We either generate the results ourselves as in the case of the dry blood spot program, or we have more time to work with families and providers on ensuring proper follow-up visits is the case in EHDI. But in CCHD, clinical follow-up needs to occur quickly often before the program even knows about the failed result. So the question is, what does follow up look like for CCHD? And when does it end? Is it simply the role of public health to collect the screening data? Do we need to go further and collect diagnostic and treatment outcomes, or do we even go further and collect post diagnostic and treatment outcomes? We need to look at models where programs screening, birth defects, maternal and child health work together to achieve the shared goals of these programs for these families.

So when it comes to education and training of healthcare providers, it also seems that this has been an area that needs attention. We continue to hear about cases where symptoms are dismissed or work up delayed because there's too much reliance on the pulse-ox screen, reminding providers of the targets of pulse oximetry screening, and the limitations of the screen is really needed to ensure that clinical vigilance continues.

There also seems to be a bit of a gap in what happens when a newborn isn't screened altogether or isn't screened appropriately. What is the recommendation for primary care providers? Should they try to do a screen? Is it too late for screening? What are the next steps in these cases? Of course, birth facility staff play a pivotal role in pulse oximetry screening. We still see misinterpretations of the algorithm. If you are changing your algorithm, if you are changing the recommendation, how will you reeducate them and really, how can we incentivize better data reporting?

For family education and needs certainly questions around understanding of the screening test and limitations. Do they know what to look for even after a past screen? Are they even being given their pulse-ox results? And do they understand maybe the integration and role of this screen with birth defects registries? The other question is, are we meeting the needs of families identified with CCHDs? Do we know what those needs are? I have an example here from the Minnesota Department of Health. I don't know if Heather [Pent 00:58:37] is on, but I think they've done a really nice job of working with families to create a guide of resources for families once they are diagnosed. Is this a model we need to look at a more national level?

So briefly I will touch on the opportunities we have in integrating other screening practices and the intersection of prenatal and clinical detection with screening detection. Many of us have probably heard about the potential of Perfusion Index in maybe helping us detect some of the defects that we don't typically pick up as well right now, but there is other work looking at integration of other screening

programs, so potentially looking at cardiovascular biomarkers in dry blood spots. So is this another where we can continue to learn and grow?

We've talked again about detection modalities. Our prenatal detection rates improving is that happening everywhere across the country? Is that happening to for everyone? And this graph here, which is taken from Children's Healthcare of Atlanta, I'm sure is probably similar across other tertiary care centers, but you can see this ongoing growth of fetal echocardiogram volume. So really, how does the role of pulse oximetry screening continue to grow with improvements in other detection modalities?

And so to end, I know we just covered a lot. We want to mention that this webinar is really just the beginning. We want to keep talking with you about this topic and have a couple more webinars planned in the series. So the next webinar we'll focus on the clinical cardiology perspective as we learn more about the targeted defects and various modes of detection. This webinar is tentatively scheduled for late fall 2021, so keep an eye out for that announcement. And then for the third webinar in this series, we hope to hear from a few programs on lessons learned and updates on CCHD projects, and this webinar will likely be held early 2022. So with that, I am happy to send back over to our moderators.

Jill Glidewell:

Thank you Amy and also to Lisa and Careema. And we will open up the line for questions. And while we're waiting on folks to unmute the lines with questions, we've had a couple of them in the chat box. We have a question that asks who is the first one in when to introduce and define the concept of CCHD? What we learned in medical school that congenital heart diseases are categorized into cyanotic right-to-left shunt and non-cyanotic left-to-right shunt. This genetic CHD also has the short-term CCHD, the critical CHD seems to be more popular in the public health fields. Lisa, did you want to take that question?

Lisa Hom:

Hi. Sure. Yeah. Thanks Jill. So I know we have several pediatric cardiologists on the line, so I'll definitely give them an opportunity to take a stab at this as well. But certainly in the literature, the definitions, we've seen them for as long as 20 years ago. I know that there's a Hoffman study that was published around 2002 that talks about the different categories of congenital heart disease. And most of the literature defines those critical forms as occurring about three out of every thousand infants. And the agreement is that they're serious and that they require intervention in the neonatal period or the first year of life.

I don't know if anyone on the line has the exact first reference in the literature, but certainly we've been seeing it for about 20 years. And then I know in England the public health community a lot of times they use different words and they have a different way of classifying those primary targets. I know many of you are familiar with Andrew Ewer's body of literature and the work that he's done in England and they call it severe congenital heart disease. So does anyone else on the line have a comment or can better answer that question?

Jill Glidewell:

Lisa, I can't better answer it, but a little bit more by way of history is the 2009 AHA statement by Bill Mailey

Lisa Hom:

Of course, yes.

Jill Glidewell:

... described screening, which was one of the first papers termed Critical Cyanotic. So both terms critical and cyanotic, and once CCHD was endorsed by HHS, and added to the RUSP, the expert panels decided to drop the term cyanotic as not all of the defects or the target list were all necessarily cyanotic. And so the sign cyanotic term was dropped and shortened to critical congenital heart disease. So I recall that from a little bit of history, but I do welcome any other pediatric cardiologists to jump on as well.

Dr. Russell Kirby:

Okay. Well, if we don't have any historians online who want to dig into the origins, which is always an interesting topic, I wanted to ask a question and I don't know if this is best for Amy, but it could be for anyone on the panel. You're talking about the need for follow-up. And I wonder if you could talk a little bit more about whose responsibility that might be and how we can best do it. The reason I'm asking is that some of the birth defect registries have the ability to capture information from hospital discharge and other sources to where they're able to actually look at longer term outcomes besides mortality. They can look at repeat surgeries or other reasons for being hospitalized for children, but it's something that we haven't really done a good job with pretty much any of the newborn screening activities in terms of really institutionalizing a comprehensive follow-up strategy. So I just wonder if you could talk a little bit more about that.

Amy Gaviglio:

Yeah. That is a big question. I'll do my best, and I think some of it will depend on how we define follow-ups. So there's certainly the follow-up that I think most people appreciate in terms of we have a failed screen, we need to find out the outcome of that screen and that surprisingly can be very difficult in and of itself. Roles and responsibilities around that I think have varied again from program to program. I think, again, those who have really integrated programs with their birth defects have been the most successful in being able to alert each other of potential failed screens and either utilize a birth defect mechanism like chart extraction to get outcome data. And then doing some of that back as well. So having birth defects say, "Hey, we have these cases that you don't have or that passed the screen."

So I would say it seems that the most successful model are those that have really integrated that work. Programs who kind of regularly share their data together and talk to each other and serve as it's a win-win for both programs, really when you do that. When we start talking about that post diagnostic follow-up, that longer-term follow-up treatment outcomes, how are they doing? You're spot on that we haven't figured that out for any of our conditions. And so there's not, I don't think a fantastic model out there. Probably best model that we see is with cystic fibrosis, but they are of course, a very well-funded and well organized organization.

So I think responsibility around that is likely, again, going to need to involve collaboration. So collaboration between public health, the specialty care centers, certainly, and also the families. So I think there are some models out there that we can look at in the chronic disease space, but I don't have a good answer for you because we haven't really defined that that anywhere. But I think now is a great time to start having that conversation because there is a lot of thought going into to longer-term registries, even across newborn screening as well. And Careema, I don't know if you have anything to add from that perspective, from APHL perspective.

Careema Yusuf:

No, I think you've covered it Amy. Currently, I think folks are looking at more disease specific registries, not long-term follow-up so maybe that's somehow we can do with CCHD. But I think to your point,

defining what we mean by follow-up, and things of that nature. I think what you just covered in the last piece slide is important to figure out. So I think it's an ongoing conversation and we're happy to hear from the folks on the ground and on what their perspectives may be.

Amy Gaviglio:

I think it's important to look at it, not state by state as well. I think that work needs to be done at a minimum at a regional level. And I think the more successful programs that we've seen, whether again, it's CF, which has done more nationally or blood disorders, which can be done regionally, that's really led to some success, but also having everyone agree upon what are the key elements to collect is also important so that we're kind of all measuring apples and apples.

Careema Yusuf:

Yeah, I think you mentioned this earlier, Amy, about the CCHD DRT the group coming together to figure that out, the set of like a common data model can be collected and then how can we move forward from there? So-

Dr. Russell Kirby:

Yeah, as that process goes on, it would be a good idea to involve somebody from the national birth defects prevention network. Cause we've done a lot of work thinking about databases and data elements for birth defects, their balance, and it's not exactly the same as what you're going to need for that, but I think that expertise might be helpful.

Amy Gaviglio:

Agreed.

Dr. Russell Kirby:

Well, we're not getting a lot of questions from my audience. I can't believe that we answered every possible question because my head is spinning with things. But if you do have a question type it in the chat, we did get a question about how to access this presentation afterwards. So looking in the chat for a link, that'll tell you where to get that later. And there also was posted a link for how you can sign up for the webinar that's happening. I guess it's Tuesday talking about vitals statistics and data integration with newborn screening.

Amy Gaviglio:

Yeah, just a touch on Jillian's comment about it being the wild west. And I do think that, that has been an interesting kind of additional variability, it's even the variability you might see within your state. So, if you don't have a specific protocol written in your statute. So Lisa showed that map of some recite and what you have to do the AAP protocol. It can be difficult to choose a protocol to recommend it in some programs or some hospitals, birth facilities may choose their own. And so I can appreciate that difficulty. I would just say if you're able to join the CCHD data response team where you want to reach out and send out an SOS for it, that would... That is definitely an option because I think we've certainly dealt with states who have hospitals using different algorithms and you're right. It becomes a bit chaotic. So kind of, how can you try to mitigate that? And I'm sorry, I think, is it one of her, were you going to say something, you're unmuted if you weren't.

Dr. Russell Kirby:

Okay. Well, Jill, do you have any other questions? Looks like we have one more. We got one just now.

Jill Glidewell:

Yeah, let's see. Chris Barnett, did I hear correctly that they need to be an increasing trend in prenatal diagnoses of CCHD? If so, is there an idea to why that's the case? I believe Amy, you touched on that if one of your last slides.

Amy Gaviglio:

Yeah. And I certainly invite Lisa or others to weigh in. And, but I think it's largely both a technology issue. So the technology is getting better. I think there are more recommendations around ways to look for CCHD in the prenatal period in potentially just because of technology advances kind of increased access to this. So I don't know if there have been actual studies looking at increases in prenatal diagnoses, but I think certainly we're seeing an increased utility or use of technologies that do detect prenatally. Lisa, if you want to add from your experience.

Lisa Hom:

Sure. Absolutely, Amy. And so I work a lot with the fetal cardiology team at national and certainly both locally and nationally, we're seeing a lot of effort in terms of improving those prenatal detection rate then that's the focus of a lot of these programs is to make sure that they're able to diagnose them early, to make sure that they're being delivered safely. Some of them can get into trouble in the immediate newborn period, as soon as they take that first breath, I'd say a lot of education in terms of sonographers and working with the MFMs and the obstetricians in terms of how to perform this filter sounds and how to... Which ones to refer. So I think the programs are getting better in terms of making sure women are getting into prenatal care. There's a lot of national effort around that.

And then as well as improving the education of sonographers, then an OBS in terms of looking at those anatomy stands and making sure women are getting them. So there's also national quality measures. And definitely there's a lot of literature, whole bodies of literature written, written around improvement of prenatal detection rates for critical congenital heart disease. But I think there are some areas of the country certainly that have published that they are almost at a hundred percent. And so those centers are, I think are few and far between, but certainly there are some places, especially big sub-urban teaching hospitals where they do have a really high level of prenatal care and a lot of expertise with fetal ultrasound where they're at or around close to a hundred percent for their local geographic area. That helps answer the question. And certainly I think it's better to get a prenatal diagnosis than to find out just prior to discharge that your infant has a serious heart defect or a critical form of congenital heart failure.

Amy Gaviglio:

Yeah. I think that's exactly a super important point, Lisa. I think it programs have kind of said. Well, we're only picking up, one, two, three or a handful of babies with the screen. And I think that's great because we are that third line and in the ideal situation is that they are picked up prenatally followed by, the next ideal is very early clinical detection. And then, you know, we're really catching those babies that have fallen through those two cracks. So it's okay that maybe aren't picking up that many. And so understanding how prenatal detection is evolving and improving, I think will help us understand our numbers by pulse-ox as well.

Dr. Russell Kirby:

Okay. So we have a couple of additional questions, one from Florida asking about how other states have integrated their follow-up efforts with birth defects to Houston, asking if we're going to include that in one of the upcoming webinars. I kind of think the answer is yes, on that. We haven't completely planned those, but we can definitely include that as one of the topics.

Amy Gaviglio:

Yeah. I think that's kind of what we were thinking for the third one, but if you have specific questions, again I encourage you to reach out to the CCHD data response team. We certainly have programs represented who have really nicely integrated their follow-up and birth defects in some interesting and unique ways. So if you want to hear kind of more one-on-one, that's also another great option.

Dr. Russell Kirby:

Right? And we have one other, I think this is more of a comment from Brianna Swartz, pointing out that there are specific guidelines for which pregnant women to refer for fetal echocardiograms and that there's no focus on assisting obese and maternal fetal medicine sonographers in identifying the signs on anatomy scans. And that comes hand in hand, of course, with the improved imaging technology that we have, but she also points out that there are some disparities in access and there definitely are. I think it probably would be correct to say that there are some equity issues that need to be addressed in terms of that as well.

Lisa Hom:

Absolutely. I think one of the biggest papers that came out in the past five years is there Quarter main paper that looked at an analysis of all infants who received well, all reporting participating centers that contributed to this society, thoracic surgeons, registry on infants who received cardiovascular surgery for CCHD. And what they found in that analysis is that certainly there were a lot of biographic trends with, I think some of the lowest prenatal detection rates at that time were being seen in Texas and some of the Southern states and that the Northeast had some higher prenatal detection rates.

But if you look at it regionally, there were no sort of regional areas that were reporting that really high numbers that we're seeing and have some specific centers who can report it in the high nineties or a hundred percent even I think the highest levels where right around 50 or 60%, when you look at it regionally, I think what we like to say is these methods of detection are not enemies of each other. They're all working towards the same goal. So like Amy said, we're really happy when infants are detected prenatally. And if that means that the yield for screening is lower, then that's better for the patients certainly.

Dr. Russell Kirby:

Because it all depends on how you calculate the deal too, if you calculate the yield, excluding those that are already prenatally diagnosed. No, but yeah, let's see, we had that-

Amy Gaviglio:

The last point is really important and I think is spot on is, and maybe that's something we need to think about with dataset. Recommendations is the importance of collecting potential geographical measures or S-E-S measures, because I think we are going to see differences in the... I'll say the utility of pulse oximetry screening probably in different areas. So the area that you described, Lisa, where they have a hundred percent prenatal detection, maybe not as not as useful, but in areas where they're not even close to that much more useful. So I really appreciate that comment and-

Dr. Russell Kirby:

That's actually important. There's a report that the March of dimes put out a year or two ago where they coined the term maternity deserts. And it's really interesting, but in almost every state, maybe not Rhode Island where there's only one county, but in most states there are sizeable areas where they're basically no obstetrical providers at all. And that has to lead to a decline in access to these kinds of obstetrical services.

And of course the geography of that varies in different places around the country. Okay. So we had a couple other things and Kathy Higgins from the Massachusetts birth defect registry pointed out, they've done some studies specifically looking at prenatal diagnosis and contribution to detection that's on. And I think somebody asked whether she had published that yet. And I didn't see an answer to that yet, but hopefully if there is, we'll certainly find it and pass it on, it looks like there is a one paper that I'm going back into the chat. There is one paper that they have and another that they're working on. So. Okay. Well, Jill, we have a few more minutes. Do you want to see if anybody has any other questions?

Jill Glidewell:

Feel free. Anyone, if you have any additional or follow up questions, you are able to unmute your mic or type into the chat, prefer to do that.

Dr. Russell Kirby:

Okay. And we got one more question and it's kind of a general question, but asking about how does prematurity, if at all affect the readings. I think they're thinking about with the pulse oximetry screening, whether the baby's extreme preterm, for example, does that affect the efficiency of the screen?

Jill Glidewell:

Can we say you've covered the NICU population a little bit, if you would like to expand on that.

Lisa Hom:

Sure. Absolutely. In a general sense, absolutely prematurity impacts the screening. A lot of times when these infants are born early, their lungs are underdeveloped. And most of them, I think maybe about a third of them are on oxygen. If not more, depending on why they're in the NICU and what their gestational ages, but certainly the under-development of their lungs can absolutely lead to lower oxygen saturations, which would render the pulse oximetry screening, much less helpful. So most of the screening protocols that I've seen require that the infant is at least, sort of early term to... we're on 35 or more weeks gestational age calculated before they're going to do a pulse oximetry screen. And they also require that the infant is not currently on oxygen when the pulse oximetry screening is happening.

Amy Gaviglio:

I believe there's a few states who have some nice kind of published NICU algorithms. I'm thinking Michigan. I know I'm trying to think Lisa, if there are others, but yeah, that generally kind of cover what you just explained in terms of waiting a bit longer and making sure they're off oxygen.

Lisa Hom:

Sure. Absolutely. There's several teams nationally that have published NICU algorithms and that have published several papers. So if anyone's interested, please feel free to email me and I can email you some other papers that have come out in the past few years.

Chris Barnett:

There was a little talk about the categorizing the timing of CCHD diagnoses. I wanted to know if there was any literature on the best convinced to categorize... Or how those are categorized and why.

Lisa Hom:

Chris, what program are you from? What state are you from? I was just curious, sorry.

Chris Barnett:

From Alaska... Alaska birth defects registry.

Lisa Hom:

Wonderful. Thanks for joining us.

Chris Barnett:

Yeah.

Lisa Hom:

So definitely, there is a lot of literature and a lot of analysis has gone on, especially Jill mentioned some of those early efforts when a lot of the professional societies were doing the literature review in terms of which algorithm to and whether the US should implement CCHD screening and they based it on one of the population-based studies coming out of Sweden. And that algorithm use the at or around 24 hours of age. And what the literature was showing and studies showed is that if you screen earlier, you may have a higher rate of false positives because some of those infants haven't fully, well, it just depends on the timing of when that infant is transitioning from fetal to newborn circulation. And so you can have a higher false positive rate if we screen prior to the 24 hours of age.

However, if you look at the literature coming out of the screening Hom births, many of those, what we call false positives are infants that don't have a critical congenital heart defect, but what we're detecting earlier, if sometimes those cases of pneumonia and sepsis and other important sort of respiratory cause for infectious cost reasons for having a low oxygen at three and so timing is definitely important. And the US in terms of the panel that recommended CCHD screening and that sort of selected the initial algorithm, did a lot of, sort of looking at the published literature to make sure that they struck a correct balance between having an acceptable, false positive rate, but while still capturing these infants in a timely safe manner. And I certainly welcome. I know there, there are several people on this call that can probably articulate that further. So please, if there are other comments that maybe happy to have them.

Chris Barnett:

That was great. Yeah. And I was also interested in how, if we're looking at the data, how, and try to define when the CCHD were diagnosed, whether it was prenatally or post-natally, or is there a standard way to categorize when each patient was diagnosed.

Dr. Russell Kirby:

Now, that is probably a more broadly of the birth defects surveillance question. At least, many of them, many of the surveillance programs actually have data elements in their databases to track that, although depending on what kind of surveillance methodology they use, it can be you more or less difficult to do,

but many programs actually cracked with the date field for the date of a diagnosis. And one can figure that out from there. Some actually have a field for, was this diagnosis made prenatally or postnatally, but it varies quite a bit across all the programs. And then again, as I... As one of our presenters mentioned also, there's also the fact that we don't have data integration between the newborn screening program and the birth defect registry in many states. And so making that connection would also be helpful as well.

Amy Gaviglio:

It may be interesting to see those eight programs that is kind of one of their charges to see how they do it and what they find. I think one of the things that we struggle with a lot in public health are the data elements that are really interesting to us are very hard to get out of EMRs are they're all over the place. And so if they find that, it's that something we need to look at and work with some standard terminology areas, USCDI, or whatever it may be to add some of that, to make some of this analysis easier. So I, that's a really good question, and now, Jill, if you have anything to add?

Jill Glidewell:

That's one thing that our collaborative group of eight funded states we're currently discussing, defining the timing that the prenatal early clinical meeting, less than 24 hours or after discharge from the birth hospitalization.

Dr. Russell Kirby:

Okay. We are running out of time. We're supposed to go to 10:30 Eastern, right? So at this point, I want to thank everybody in the audience for your participation. It's been great to have all the dialogue and feedback, and I want to thank each of our presenters. This presentation was recorded. So it will be available later on a website and keep your eyes and ears open for when we schedule the next webinar. We're hoping to do that probably before the end of the year and then have a third one probably after new year's. So we definitely, and we also welcome your feedback. If there are major areas that you want to have more detail about, or if there are things that we didn't cover, that would be important. Definitely let us know, and we'll try to get the information and share it with everybody. Okay. Well, I guess we can conclude and have a good rest of the day.

Lisa Hom:

Thanks everyone.