What’s New in Texas – Newborn Screening Program - Pediatric Grand Rounds-11-7-2025-Meeting Recording

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1h 3m 52s

Calderon, Delia started transcription

Gong, Alice K 0:04

OK.

Ranch, Daniel 0:35

All right, it's 7:30, so let's get started. Good morning, everybody, and welcome to UT Health San Antonio Pediatric Grand Rounds. As a reminder, as you log on, please mute your devices. Also the.

Grand Rounds attendance code will be placed in the chat periodically, so you can just check there for for attendance. Otherwise, it is my absolute great pleasure to introduce our speaker for today, Doctor Alice Gong, who actually needs no introduction whatsoever, but we're going to do it anyway.

Doctor Gong obtained her medical degree from the University of Mississippi School of Medicine and then pediatrics residency initially at UTMB Galveston. She then shuffled off to Buffalo with her new husband to finish her pediatric residency at SUNY Buffalo.

Doctor Gong then joined the UT Health San Antonio faculty in 1985 after completing her neonatal perinatal fellowship at SUNY Buffalo. So and she's been here and has risen through the ranks to professor and beyond while she, with her bare hands, helped build this department to what it is today.

Her passions in neonatology include nutritional support of premature infants to include breastfeeding, respiratory distress syndrome and surfactant replacement, retinopia, prematurity, and follow through care of NICU survivors. Her most recent work has been in improving family support of NICU babies to improve their outcomes.

She has served on the Texas Newborn Screening Advisory Committee since its establishment in 2010. She's currently part-time and plans to retire next year. We'll see about that. And she's most proud of her three children and four grandchildren.

So Doctor Gong, you have the floor.

Gong, Alice K 2:15

Thank you, Doctor Ranch to minimize this thing, so.
So you won't have to see it. OK. Thank you very much for this invitation. And newborn screening has also been one of my passions and it's been there's been a lot of changes in newborn screening. So I hope we all learn something from this.
I'm trying to advance my slide and it's not working. Oh, there you go. So what I'm going to do if I can get rid of this thing, which doesn't want to go away. Are you seeing this on your screen too?
Anybody?

Ranch, Daniel 2:55

Dr. Gong, we see your slides perfectly.

Gong, Alice K 2:57

Oh, you do. You don't have the this thing that I can't get rid of. OK, so I want to go over the purpose and principles of newborn screening program because it is a cornerstone to Pediatrics summarize.

Ranch, Daniel 3:00

We don't have the extra notes or anything. Yeah, yeah.

Gong, Alice K 3:12

National policies that involve newborn screening and then characterize the uniqueness of newborn screening program in Texas. And hopefully you can kind of recognize some of the conditions because there are a lot of conditions. I'm going to go through it somewhat cursory.
Because each one of the conditions wants its own individual grand rounds.
OK, So what is newborn screening? It is the practice of testing all newborns in the US, and it is all newborns in the first days of life for certain disorders and conditions that may either keep them from living or hinder normal development.
And it is required in every state and US territory in their public health system, and it should be performed before the baby either leaves the hospital or the birthing center. The conditions in newborn screening typically are those that have no symptoms at birth, so the baby looks perfectly.

Normal, but can cause significant morbidity and mortality if not detected and treated early or quickly. This is one of the tenets of newborn screening that you need to have something that you can recognize before there are symptoms and that you can treat and prevent damage. And it has been.
One of the most successful public health interventions in the 21st century, mostly because a lot of what we do in the 21st century was not done in the 20th century. Auto newborn screening started then. OK, so some facts. There are about 4 million births in the US, so.

Brooks, Edward G 4:42
Mhm.

Gong, Alice K 4:56
One in 178 found to have some kind of devastating condition through newborn screening. And in Texas, newborn screening started in 1963 with the Guthrie test, and that's how we got to filter paper.
So all the states, District of Columbia, Columbia, Puerto Rico screen now for 31 of the 38 recommended disorders. And I'll go over how you get to recommend it. So annually, about 20,000 newborns in the US benefit from this early detection and life-saving treatment.
Most babies with serious but treatable conditions discovered by newborn screening actually grow up healthy with expected development. So in terms of the federal government.
The first law was passed in 2008, but prior to that it was actually recognized that states were doing things differently. And so it actually was 1 congressman who found out that his sister who had a baby in another state got more testing than his baby did, which started.
Task force and studies and so a law was passed called the Newborn Screening Saves Life Act that says there is going to be a national newborn screening guidelines to support states so that they can get more to more uniform screening.
And prior to the pass of that law, there were only about 10 states that were doing most of the recommended screenings that had been set up by the genetics group.
And then after the law passed, we have now gotten to almost all the states.
That law lasted until 2014 and it was reauthorized. And when it was reauthorized, there were things in there that improved and expanded state newborn programs.

Address the need for education and then having some federal policy for protection and then set up standards and surveillance efforts. That was to expire in 2019. In 2019, the House passed it, but the Senate did not address it and so.
It was it kind of been waiting for the Senate to approve it, but it did not. So in 2021, the House we authorized again giving until 2026 for the Senate to pass it, and it did not.
OK, So what the authorization would have done was that there would have been HRSA grants to help states expand and improve their programs, provide education for parents and health care providers and improve follow-up care and also allow for the CDC to do.
Prevention newborn screening quality assurance programs to make sure that testing is accurate. It started a program called Hunter Kelly, a newborn screening program that will fund research so they can identify new treatments and develop new screening technologies. It set up the Advisory Committee on Heritable Disorders.
In newborns and children. So this is the act, which is very difficult to say, but that's the committee that goes through the process of looking at conditions through a lot of.
Vetting that includes not only the experts that are doing the studies, but also populations affected. So parents of children that may have had that disease as well as other stakeholders and they are the ones that can allow news.
Testing to go to the Recommended Uniform Screening Panel at the RUSP and they also would have allowed for the National Academy of Science to develop policy recommendation on how to modernize the system as new equipment and technology becomes available.
So that act did expire and so in 2014 and what happened was that the Advisory Committee on Heritable Disorders in Newborns became a discretional committee under the Health and Human Services to continue to provide.
That function and unfortunately this committee was disbanded in in April of this year.
So although this federal legislation, actually what who is responsible for newborn screening is the individual states. And so Texas has two policy statements and they're in different.
Places, unfortunately. So Health and Safety Code Chapter 33 actually covers all of the newborn screening that is done by the lab. Legislation has been passed that the lab can add new conditions that are under us as funding allows.
And CCHD screening, which is critical congenital heart disease screening, was added

to that area. The hearing screening actually went under a different law in Chapter 36. That's something that is going to be there and we can't change it. OK, So what is the recommended?
Uniform Screening Panel. It was established to standardize the list of disorders that are supported by the Advisory Committee on Heritable Disorders in Newborns and Children. They recommend that a condition.
Be added to the Secretary of Health and Human Services, who then signs off and then it becomes part of the rest. So the rest is still there. But what happened is that our states. But remember the states decide for themselves what conditions to add and as of today.
The Texas New World Panel contain has all the conditions with the exception of guanidino acetate, methyltransferase deficiency or GAMP, which I'm going to refer to because it's a lot easier to say that implementation is planned for either December or early spring of 2020.
So some history about newborn screening. So as I've mentioned before, in 1963, Texas started PKU screening. That is why a lot of people still refer to newborn screening as PKU, although PKU is only one of many conditions that is funded. So for 30.
35 years Texas funded newborn screening. A fee was not instituted until 1998 when about that time around the country there was developed tandem mass spectrometry which could then I think someone's got.

Perlman, Jeremy S 11:54
Now that's all.
Uh.
Yeah, that's good. Thanks.

Gong, Alice K 12:06
That's OK. So what happened was Texas was still screening for the same five conditions. And so through some advocacy work with Texas Pediatric Society and Texas Medical Association, a task force was.
Form to to look into expansion of newborn screening. That task force reviewed the literature and recommended expansion and actually it was not and then asked for legislation. So House Bill 790 in 2005 actually expanded.
Screening stating that tandem mass spectrometry is is what is needed. So the that's

what happened with that law also came about the Newborn Screening Advisory Committee.
OK, So what is unique about Texas? In addition to expanding screening, it became apparent that the other blood spots that have been around since 1963 were being kept and there were ways that you could do testing.
If you go through the IRB at the health and at the state health department, people got upset about that because the government had their kids DNA. Therefore there was a lawsuit filed against the newborn screening of.
Program In order to avert the lawsuit, this program may recommendations to the state legislature. So House Bill 411 in 2011 actually addressed the issue of the blood spots.
So basically, if you if parents consent, the specimens can actually be stored up to 25 years and can be used for external research if it passes the IRB and it meets the approved requirements that are stated in the legislature if the parents don't give consent.
Specimens are stored up to two years, not used for external research, but maybe to use some for quality measures to kind of make sure things are that they meet the standards.
So because of that law, we new forms were made, collection kits would change. So storage of the newborn blood spots is an up in process and parents have to approve it. So what happens with the kids that come out is this form where parents this should be given to the.
Mother when they're doing the first newborn screen and they can check yes or no or OK means the blood spot will be kept. No means I don't want the blood spot kept. It will be destroyed after two years.
They can give that back to the staff and that can be sent to the state or they can mail it in because Texas does 2 screens. If the first one says yes, but the second one says no, it will not be kept. If the first one said no, but the second one says OK, it will be kept so.
Those are the conditions. So now newborn screening itself is an opt out. In other words, it is state mandated and the only way, according to statute, that you can refuse newborn screening is if you have a religious objection.
So what? And there is a form that is that can be found on this link to get to the that form. So the process that is recommended to pediatricians and people who attend the birth of the baby.

Who are responsible that newborn screening be done is that if a parent does not want newborn screening, you need to be able to discuss the general purpose and benefits of newborn screening, answer questions, provide some printed information, which I'll show you on the next slide, and then explain that the only.
Legal reason to refuse is if the test conflicts with their religious practice of an organized Church of which they're members that is written in statue and then show a video which talks about how wonderful newborn screening is. These are some of the things that you can find online that you can print.
And share with families. So there is a form and you should download it if you have a parent that says no, I do not want this done. And basically you fill out the form and they have to check that I have heard the benefits. I know I can only refuse this because it's against my religion. I don't want my.
Test my baby's tested and you're supposed to give them that copy because they're supposed to show that to the baby's doctor so the doctor will know the baby did not get a newborn screen. And then the bottom part is supposed to be filled out by the facility and sent to DSHS.
I don't think the system works that well because we don't get very many of these. OK, So what is what does Texas do? We screen for 59 blood disorders. It will be 60 as soon as GAMP is added. And then there's two point of care. One is hearing and.
Is critical congenital heart defects. The blood spot tests are done on filter paper, the first one with 24 to 48 hours of life. This is true for every baby, so even the NICU babies get it within 24 to 48 hours if a baby a NICU baby was transfused.
Before that, there's a caveat to send it and then to remember to send another one after you've passed three months from the last transfusion 7 to 14 days. Again, same for NICU babies. And the reason we do this is because it maximized detection and actually.
A good portion of the congenital hypothyroidism is found on the second screen. Texas is one of 10 states that do a second screen, and there was a comparison between states that do first screens and second screens, and the states that do second screens continue to find.
Conditions beyond the first screen. The cost of the kit right now is $60.63. Due to the fact that we're adding a lot of new conditions, the test will go up to $94.81.
There are two different forms. There's a Medicaid CHIP and charity form and then there's an insurance and self-pay form. The Medicaid CHIP charity forms are provided free and the insurance self-pay have to be the kids have to be paid for.

OK, I'm going to go through all the disorders in a very cursory order. First is the amino acid disorders. This is where an enzyme is defect is not there or not enough there and so it causes build up of abnormal amino acids, the classic.
Is your PKU. That is the all the conditions here are done under the tandem mass spectrometry. The red ones are those that are core condition. Texas also screens for the secondary conditions that are in black and that is going to be the key to the conditions. So amino acid.
Disorders. The next one is fatty acid oxidation disorders. These are disorders where enzymes are not there to breakdown fatty acids and therefore can cause problems. The major one that you may know about is MCAT, which.
It's very important to pick up early because when babies are not fed regularly and they don't have fuel, they don't have, they can't generate energy and that's one of the reasons why they get into a lot of trouble early on.
Organic acid conditions are those where enzymes don't break up certain amino acids, organic acids and there are non core conditions. Actually we NICU actually had a case of MMA recently.
OK, so endocrine disorders, I don't have to kind of go over those because usually we all know about congenital adrenal hyperplasia as well as primary congenital hypothyroidism. These are two conditions that are more frequently picked up on the second screen.
And in the hemoglobin disorders, in addition to the beta, SD disease and sickle cell anemia, the state also does screening for all the hemoglobinopathies. Lysosomal storage disease just got added in August and I'm going to go through each one of. These, but there are Pompe, MPS, and infantile cat krabpe disease, other disorders of which I'm going to go through. Some of them are X-linked adrenal leukodystrophy, biotinidase deficiency, classic galactosemia, cystic fibrosis.
And SCID as well as SMA. In addition, because you're screening for SCID, you get the T-cell related lymphocyte deficiencies, which is not a core condition. And then we have two point of care, which is hearing loss and critical congenital heart disease.
OK, so kind of cursory, but I think these are the conditions that you need to know what the state does. So as you all know, cystic fibrosis is an autosomal multisystem disease that affects quite a few US newborns.
Is caused by genetic mutations involving the cystic fibrosis transmembrane regulator protein, and because of that you end up with all the different issues that we see systemically.

What Texas does is it looks at immunoreactive trypsinogen, which is the IRT. If the IRT is abnormal in the 1st and the 2nd screen, then a DNA approach is used and Texas screens for 46 of the mutations.
Picking out those that are more common in the Hispanic population. Obviously there are more than those gene mutations that can cause cystic fibrosis. So unless you have that DNA positive for the gene if they have abnormal IRT's.
Sweat chloride is still the gold standard and it's very nice cause see the CF centers in the state actually do a lot to help with the follow through on on this test particularly.
So what happens when you don't get a second screen?
And on a baby. So CF was what required the state to try to connect the two screens, which is somewhat difficult because names change, providers change, so many things change between the 1st and 2nd screen, but they're actually able to.
Do that about 80 to 90% of the time, connect the two. So if you get a first screen and the IRT is abnormal, but you didn't get a second screen, they will go ahead and do a DNA approach. Do the DNA testing. If you only get a second screen, they will still go ahead and do DNA.
And there are times when we have reason to suspect because of in utero situations, we can actually call the state and tell them to just go to DNA. That's cystic fibrosis.
Severe combining immunodeficiency screening is quite neat. What it does is it does.
A PCR looking for something called tracks. Tracks are actually formed when lymphocytes are being formed and what they're doing is that the DNA is has to get rid of this piece of material it doesn't need.
When it happens, those little circles are called tracks and there's a surrogate marker for marker for what the thymate activity has been recently. So if it's low on both screens, so sometimes we get a low one on first screen if there's nothing that makes you think there's something.
Something wrong. You can wait for the second screen and when that happens then the the tests are done. The baby needs flow cytometry looking at T cells and B cells and NK cells so you can figure out the subtypes and then there's genetic testing to confirm whatever mutation. So if you.
Have abnormal first and second screen, you need to refer to a clinical immunologist.
SCID is that condition that prevents with infections very early in life and other autoimmune issues like rashes and liver dysfunction, hepatosplenomeric.
In diarrhea, very difficult to diagnose in the past. It is more common in males because if it's an IL 2 receptor gamma chain deficiency that is X-link, it can detect

syndromic immunodeficiencies like the George syndrome.
DNA repair disorders, metabolic disorders and even extreme premature babies whose thymus may not be working so well. Any severe congenital infections or some other malformations. So.
It is a broad screen, but it can be narrowed down. The treatment is getting a stem cell transplant to reconstitute the immune system with normal immune cells, and the outcome is amazing if you can do it before you ever have infection.
On OK, so XALD is where the very long chain fatty acids, which is your carbon 24 and 26, cannot be broken down South. You get this buildup of VLCFA and that disrupts myelin, causes nerve conduction issues, affects the.
Nervous system and the adrenal glands. It is again more common in males, although some women carriers do have symptoms in adulthood. So what the screen does is that we pick up the.
The C2426 and therefore you need to get very long chain fatty acid levels as well as your ABCD1 gene because that is a common gene that causes XALD. There are three forms and we can identify. We can't tell you which form, but the state can.
Tell you that if you don't have the childhood cerebral, which is the very early onset, develop adrenal myelone neuropathy and Addison's disease. So the positive screen, the positive cases are followed by neurologists and there have been a number picked up. Sell Wagers is one of the.
Conditions that can present with abnormal screen.
SMA, SMA is autosomal recessive and it's degeneration of alpha motor neurons in the spinal cord and so you that leads to muscular atrophy. These kids are not typically seen in the newborn period. It is relatively common.
One in 50 Americans carry the gene. So what happens is that you can have mutations of your survival motor neuron one on exon 7 that produces the SMN protein that is needed for the nerves to.
Control muscles. If you don't get an S, if your SMN one doesn't work, you have an SMN 2, which is your backup gene, and the more copy numbers you have, the less severe your disease is.
So the screen identifies the SMN 1 deletion and also looks at the number of SMN 2 copy numbers that you have. And if you have two or three copies of the SMN 2 only, then you can you do qualify to get.
The therapy with that is available. So remember there are four types of SMA, with type one being the most severe that you weren't a Hoffman. These kids are typically

present pretty early with muscle weakness, trouble breathing, swallowing, coughing. Uh.
I don't know if the therapies actually work for type one. Type 2 is where it things a delay, so baby looks good but then starts having delay milestones or not developing any milestones. These kids typically cannot can maybe could sit up but cannot walk. Type 3 is Kugelberg.
We Landa presents a little bit later. This is the juvenile form. These kids can walk, but they have mobility issues. And then type 4 is the type that presents in adulthood with mild motor impairment and actually in less than a year after implementation of SM1 SMA.
Screening we have the state identify 16 cases so treatment is available. What was first available was nusin nursin which is an what it does is it increases the ability of the. SMA protein to last longer. It is given through a spinal tab. It is quite expensive and has to be repeated. So Gensma or Anna anyway is the first gene therapy that's available for children.
It uses an adenovirus vector to deliver a fully functional copy of the SMN gene into the target motor neuron cells. It is given as a one time IV thing. It is quite expensive, but it is FDA approved.
And then finally there is an oral drug that is available that is an SMN 2 directed RNA splicer modifier. With an open label trial, they were able to have some improvements. For late onset, the primary endpoint was that they have increased in this FM MFM 32 and they were able to achieve that. So it is quite expensive.
Based on the patient's weight and has to be given more than once. OK, so on to the new glycogen storage diseases that I mean the lysosomal storage diseases. These are conditions where you're missing something in your lysosomes therefore.
You can't get rid of things that you should be able to get rid of. Those Pompe was the first lysosomal storage disease that made it to the roughs. This happened in 2015, but because of the need for special equipment to do lysosomal storage to good.
Disorders. It took the state quite a while and so all the lysosomal storage disorders came on board now. So this one is absent alpha-glucosaminase or GAAA. There are three forms, the infantile form which presents with myopathy, hypotonia and.
Failure to thrive, poor feeding, respiratory distress, etcetera. There is the non-classic infantile onset, which is delay until age 1 when you start getting delayed motor development, weakness, respiratory distress. And then there's the late onset where

you develop progressive.
Weakness. There is enzyme replacement therapy to increase the GAA and it's very important when we diagnose this to get this going. OK, the next one is sorry, I have something in my way.
OK, it's MPS one which is missing the alpha L Iuandidase or the IUDA. It is a progressive multisystem disorder while using tandem spectrometry. We can detect the low IUDA enzyme activity and then how to diagnose it.
You have to measure activities in either the leukocytes or skin fibroblasts. You cannot predict the phenotype, and there are many genetic issues, but you can get a stem cell transplant to help that to so that you can produce the endogenous.
Enzyme or you can get enzyme replacement. Although enzyme replacement for MPS one does not cross the blood brain barrier.
So Hunter syndrome or M PS2 is X-linked. It is missing glycosaminoglycan. And so to diagnose it, you want to find that this enzyme is deficient in white cells, the fibroblasts or plasma. There's an early progressive form.
Which gives you pretty bad things where you're more likely to die in the 1st and 2nd decade. There's a slower progressive form. These kids actually have distinctive features and and there is enzyme replacement therapy.
And getting stem cell transplant can help in this condition. The last is the one that's more important that we diagnose early because we're only looking at infant infantile onset Krabbe disease.
This this condition affects the myelin, so you get a rapid decline in the nervous system. It is the galactolipids that build up. So basically the the screen is cyclosine levels.
And you may get a report that it's borderline or abnormal. If it's abnormal, you should be able to get results within four days of the example. And the reason is that this kids need to get into stem cell transplant within 30 days of life. So it's very important that we pick these up.
Expected one to two cases of in Texas. We have three places in Texas that will serve as the Krabbe Referral Center and notice that Doctor Odom at CHRISTUS Health in San Antonio is one of the sites.
OK, last kid to come to the plate will be GAMP. It is very rare. There's about 110 cases worldwide. It is due to mutation in the gene and that is essential to create creatine so.
When you don't have enough creatine, you have higher of the granulino acetate,

which leads to serious brain and muscle symptoms. So there are a lot of cognitive impairment, language, behavior problems, seizures, muscle, poor muscle.
Tone, poor muscle skills. If the symptoms don't develop until after the maternal creatine is used up, so around 3 months and early diagnosis is very important. Treatment is actually giving creatine and getting a diet low in arginine.
OK, so Texas does have a couple of tests that are not in the RUS. Unfortunately the CMV legislation passed in 2023 when we still had the RUS. So in thinking that RUS will pick up CMV because.
During that time, the way to detect CMV, the best way is through saliva. But there are tests now that can pick up CMV in the blood spots. Not as effective as saliva, but it would be a good way to pick up congenital CMV knowing that a lot of congenital CMV presents with hearing issues.
Issues. That legislation says that if a baby doesn't pass the initial hearing screen in the hospital, they need to have their CMV tested. They did recognize that not all places can produce the or perform a congenital to CMV test, which is a saliva test. But they said that you need to refer. Now there's no enforcement. This is just what the law says. And sorry, you got to go back up. OK. And so, so basically there's no tracking. It's just this is we have a law.
OK, the Shane muscular dystrophy, which was another condition that the rest was considering before everything got the span it got. We did get a law SB1044. There's a lot of families pushing for this. The law that say that implementation is not until new lab spaces.
Built, which is not slated to happen until 20/26/27. They will need new equipment and that equipment cannot be funded except through legislation. So the families that are they pushing for this are already gearing up to.
Get legislatures to provide funding at the next legislative session. OK, so lots of new conditions and I know you won't remember them all, but hope you remember some of them. Newborn screening is a system. It has to be the babies at the center, but many, many different things are affected by.
And just remember that it takes a big village to make it all work and to make it work as well as it does. And in Texas, we have a very robust newborn screening system.
OK, so how is it done? The blood spot, the filter paper. It's very interesting. They just did a survey of.
Providers in the state and only 60% knew that the first blood spot was done at 24 to 48 hours.

So, so if you go home before 24 hours, you just do it as soon as you can. I mean as late as you can before the discharge and the second one is 7 to 14 days.
The sample is by the legislation to be collected by the healthcare provider or designee. So if you're in a hospital, you can designate the lab to do it. Texas, we went through when we were going through like back in the trying to figure out legislation, looked at a lot of laboratories.
And to see if they would just use one laboratory. And in fact, the state lab in Texas performed is the largest lab that does newborn screening in the world.
And that's because California, that has more births than Texas, has five different labs as opposed to one. So we have one centralized lab that does all the testing, lots of positive benefits and some issues there. Hearing is to be done 24 hours.
Or before discharge. And that is because in that first 24 hours there's still gunk in the ears, and so it's a little harder. There are two different tests that are acceptable OAE or otoracoustic emissions or auditory brainstem response. I'll go through those.
Hearing what you want is to screen babies by one month. So those babies that are born in areas like birthing centers that don't have screening technology, they need to refer those babies to some place that could do that diagnosis and that is by pediatric audiologist by three months and.
Intervention by 6 months and that is getting them into hearing places where they can get hearing aids, et cetera. Critical congenital heart disease is again 24 hours or before discharge.
And that's because you want the ductors to close. It is done by pulse osymetry looking at pre and post ductal sets. OK, so this is a report card for Texas newborn screening. This is for the year 2023 and the reason it's 2023 is because takes time to.
Gather all this data and you know that there were 3392,000 etcetera screens. That is probably the birth around the numbers of birth. There's probably some missing there. We do not know if every baby in.
The state of Texas is screened because it is not tied to birth certificates. It is. There's a project that's being worked on, so we will know if every baby is screened. If you double that number, it's going to be about 25,000 more than the total number of specimens received.
And that's because there are there's drop off between this first and second screen. There's a lot of things about when they collect and so forth. The unsat rate is .9%, which is not that great and that's something the state is working on, but this is their report card.

There are a lot of room for improvement. OK. So of those in there were, there were some, there were quite a number of resumed positives, about 18,000, which is true, but then there were about 2000 critical ones and.
Thus far, there's about 1151 diagnosed cases, and that's because there's still a few cases that are still being worked out. But that's typically what we see about 1000 diagnosed cases. This is from the blood spot. It's not counting hearing or critical congenital heart disease.
OK. So there is clinical care coordination that's very much part of the system and it's to make sure that anybody who has an abnormal screen gets walked through the process. So the CCC staff will notify medical providers of results and provide recommendations.
Recommendations for follow up testing and other intervention. So as I said, over 1000 babies are diagnosed, so there's all of these babies are screening. The staff do work six days a week, so including Saturdays, so that if you get a call.
Listen to them. OK, so let's go into hearing. Hearing is a mandated test for Texas and there is only there. So you have to opt out and there are refusal forms at the state lab.
We want to make sure that babies with hearing loss are identified as early as possible. It's one of the more common conditions affecting about one in two to 400 babies. The idea is to make sure they get appropriate interventions. Babies who identify early have normal neurodevelopment.
So there's two screenings, the OAE and the ABR. OAE is the otoacoustic emissions. The baby gets a probe in the ear and then a sound is produced. And what you do is you can reflect on the peripheral auditory system to the cochlea outer.
So if you have cochlear hearing loss, you may or may not get a positive fundus because if there's middle ear fluid or external canal issues, you may not get a good screen. It is the more widespread because it is.
Easier to use. If it's initially abnormal, you do need to repeat it. And so it is automated with algorithms on computers, so it actually can be done by people with not a lot of training. Important thing to remember about OAE is it does not.
Identify auditory neuropathy looking at the auditory brainstem response and I hope you see this picture, but this little probe here and a little probe in the and it records the neural activity in the cochlear, auditory nerve and brainstem response to auditory.
Stimuli. So it does reflect the status of the 8th nerve. So you can identify auditory

neuropathy. Baby has to be asleep and it is automated. So if they're abnormal in in this test and they do need to go to an audiologist for testing.
Places that take care of NICUs need to have the ABR because NICU babies tend to have more auditory neuropathy.
So we had more legislation regarding newborns hearing screening in the 20/20/19 legislative session. Senate Bill SB 1404 says that we now have to have consent to document that any in.
Any hearing and intervention outcomes can be shared with the state Teddy program and then if necessary that information can be shared with the school for the deaf and there is a consent form. If you don't have a consent form, you can still do the screen you can put into the.
Teddy system that the screen was done, but you cannot put the results of OK. So one thing that's very important to understand about early childhood hearing loss is that 50% have some kind of genetic abnormality and most of those are non syndromic and they are.
Mostly autosomal recessive. There are genes that have been identified. So kids who have congenital deafness need to have their genetics tested. Another 40% are caused by congenital CMV, and we've already talked about CMV and its issues.
PADI is the state hearing thing. It is a web-based hearing database. It tracks and it tracks intervention. It also offers certification of newborn hearing screening programs. We do a really good job with screening of 98% of newborn.
Are screened. Again, the incidence of hearing loss is one to two per thousand. School age children are more prevalent because there's ongoing hearing loss much more prevalent in the NICU population.
Whether the ABR is recommended for screening, they do have auditory neuropathy as well as sensory neural hearing loss. But because babies are born so early, frequently following the Joint Commission on Infant Hearing and the Teddy goes, which is screened by one month, diagnosed by 6 months intervention.
By 6 diagnosis by three months and intervention by six months is not usually followed. There is data that babies as early as 34 weeks gestation can be screened.
So resources for newborn hearing, there's an infanthearing.org as well as and they do go into what all the states do and there's babyhearing.org that are good sources of information.
Critical congenital heart disease screening came about in the state around 2015.
They with legislation incidence is about two per thousand. We screen for hypoxia

using pre and post ductal SAT strip monitors.
Things that are left side outflow obstruction may not be picked up with this because the ductus is still open. There is ongoing research about using perfusion in index as opposed to looking at the oxygen saturation.
Actually, in about 2022, there was convened of the experts in CCHD screening as well as other stakeholders and families to look at whether the algorithm that was suggested a decade earlier was actually.
Really what we needed to do with the all the updated research that had been done. And so that group recommended a new algorithm and then also recommended instead of having seven core conditions that all of the congenital heart.
Heart diseases should be listed as core conditions and that secondary conditions are other things that can cause hypoxia, such as hemoglobinopathy, hypothermia, infections, lung disease. Whoops, I don't know why that.
Popped up or other things like that, anything that can cause hypoxia. And so it took a while, but AAP eventually came on board and so these are the new recommendations and what's new is that instead of having.
Greater than 95 as pass in either the right hand or foot, which is your pre and postdectal. You have to have both and if you are less than 90 in either one then it is a fail and you need to investigate.
And then if it's between, if it is between that 90 or 95 or if you have a difference of greater than 3%, then you retest. Instead of retesting twice, you only retest once and if you retest once and it's still in that in between range, it's considered.
Consider a fail, and the reason is that it was better to go ahead and test those babies to see if they have critical congenital heart defects instead of waiting longer.
OK, so the reporting that Texas had the unlike hearing where there is tracking and and so forth, there was no tracking built into the legislation. So what DSHS did was to learn what the confirmed diagnosis was and so.
They created a reporting form and you were supposed to report it. Well, that didn't work very well. So despite what we know from birth defects registry that we probably have about 700 cases a year.
In the eight years or 7 1/2 years since we have had tracking only about 1300 cases were reporting. So that type of reporting did not work, but because it is an unfunded mandate.
What the state did do was make a better form that can be submitted electronically. So there's a new e-mail address and you can submit it electronically through the e-

mail, making it easier because in the past you had to print out the form, fill out the form.
And fax it, which did not work well.
OK, so a lot of information to give you. Can you remember it all? Just remember every condition that is a part of screening has what we call act sheets that you can access and fact sheets. Some of the fact sheets have been retired. People are working on them if you just know the disease.
And you put it into your search engine, you can find this information and the act sheets will tell you what to do and it does it in language that parents can understand.
Very important to have family support. What good is it if you diagnose a condition and people can't take care of their children?
So there is a program that the children with special health care needs all the kids that have metabolic conditions are referred to them and if you don't, if you can meet the financial requirements.
Then you can participate and so it you get your care provided. CF patients are eligible for CSHCN for life.
If you don't have Medicaid or CSHS, CN or CHIP or any insurance at all, the state newborn screening program will provide food, medications, vitamins, testing and follow-up care at no cost or reduced cost. But you have to be a Texas resident. You have to be less than 350% of federal.
Part of poverty level and priorities given to children two years and younger. So what's in the future? Whole genome sequencing. The history is we had the CRISPR gene technique.
A program was has been done to show that genome sequencing is can work in newborn screening. We do know that there's gene therapy that has been approved.
New York State has done a program that was funded by NICHD.
So there are some target genes that may be done. There has been a program called Newborn Screening Translation Research Network that up that try to establish a.
A program to kind of look into the possibility of whole genome sequencing. There's a new program called Beacons that has been funded by NIH. It says pilot 5 to 10 states, but what was announced was about.
4 sites that will will look into how you can do this program and I think a picture is worth 1000 words. So this is Jessica who said.
I will never forget the words. My son tested positive for SMA on his newborn screening without treatment. Most babies do not live past 8 to 18 months. I sat there

in disbelief. There are currently 3 treatments. They chose the.
Gene therapy and basically, you know, they know that there's a lot of unknown still, but the baby is thriving. And so there's a lot that we still have to learn. And but the nice thing is we have conquered a lot of diseases that were very.
Frustrating when we didn't have this therapy. So these are my references and I'm open for questions. I think I have 4 minutes.

56:29
No.

Ranch, Daniel 56:31
Yeah, you got some time, Doctor Dong, but thank you very much for that fantastic, very comprehensive review. Some of those slides definitely elicited some PTSD symptoms for my board's review days. But yes, it's open up for questions. Please feel free to place in the chat or raise your hand. There's a couple there right now, Doctor Williams asked.
How is screening handled for home births or those births not at a hospital or birthing center?

Gong, Alice K 56:56
Thank you, Doctor Williams, for that question. It is required. So they're supposed to take their child to some place that can do the screening. OK, it could be their pediatrician, it could be. I mean that has to that is a part of the law that they have to do.
Now, do they do it because we don't tie to birth certificates? We don't know. And that's one of the things the advisory committee has worked on for a decade. Like we need to know like who's not screened. If you think about the numbers of not people that may not have gotten the second screen, which is about 25,000.
That's enough to have several conditions that could have been picked up.
Oh, thank you, Doctor. Donna Beth. Now that Texas is now up to 60 mutations.
Thank you. I didn't. I guess I should have done that.

Ranch, Daniel 57:44
Yes.
Mm.

Arandes, Michelle 57:54

Doctor Gong, this is Michelle at on this. Thank you for a a really informative talk and and really helpful kind of update. I just, you know, as a as a person who works in the newborn nursery, it used to be a rare event to have somebody refuse a newborn screen. I remember having to, you know, dig through to find the form and all those things.

It is literally been at least once every week that I've been on service for the past six or seven times I've been on service, so it is increasingly problematic. So I just caution pediatricians to be explicit in their query of patients who are coming to their office and not make assumptions that a newborn screen has been done, especially if there's any other.

Gong, Alice K 58:21

Oh no, I'm so sorry.

Arandes, Michelle 58:34

You know, sort of standard of care discussions that are, you know, prefer, you know, parents are preferring not to do certain things. It is increasingly prevalent despite a tremendous amount of education and time spent, you know, trying to provide that insight to families. So just it's it's an increasing challenge which is.

It's a little scary because Texas does do a wonderful job of offering so much potential, you know, care in in in identifying those diseases early. So, but thank you for continuing to fight the fight at the state level.

Gong, Alice K 59:09

Michelle, have you shown them the video before to and they still say no 'cause that's a great video.

Arandes, Michelle 59:14

Yeah, I I will, yeah. And I will say I think the efforts in the the population I'm talking about is really a population that probably has to be hit harder up front, way up front in the in the OB part of their care.

Gong, Alice K 59:21

Yeah.
Yeah.

Arandes, Michelle 59:29
Because by the time they reach us, several of their birth plan goals have not been met because they never intended to be in the hospital. And so we're fighting an uphill battle. And so a lot of times, you know, we're trying really hard to make sure they get their vitamin K and, you know, minimal things. I've had babies leave it. End up coming in after 75 hours of Labor at home who want to leave at six hours of life, you know. So it is, it is increasingly, you know, you don't think about it, but it is a little bit of the Wild, Wild West in those settings.

Gong, Alice K 1:00:04
You should talk to Tiffany McKee Garrett because she's on the newborn screening committee and she does newborn for Texas Children's, and she's been complaining about these things for quite a long time because they've had that population quite a bit.

Arandes, Michelle 1:00:12
Mhm.
Yeah, but but thank you.

Gong, Alice K 1:00:20
Yeah, you're welcome, Jane. Doctor Lynch.

JL

Jane Lynch 1:00:25
Hey, that was great. I have a question. Do you know what's screened in Mexico? Do we have an easy list to look at to know what gets missed?

Gong, Alice K 1:00:38
There are a lot of countries that don't do any newborn screening and there was a time when I was working with some people that were like going to countries to say that you need to do screening and they were just trying to do basic screening, so. I have no idea what the law is in Mexico. The assumption would be that they didn't get any screening. And you do know that if you get a baby they they that used to be

you could screen up to a year. You could just go ahead and send the screening when when you see them if.

Jane Lynch 1:01:06
Yeah.


Gong, Alice K 1:01:16
I think they're going to relax it even more for like adopted babies and stuff like that, so you can send in the screen.

Jane Lynch 1:01:27
That's great. I I think, I think it's maybe a private pay thing in some areas, but we always worry when kids have been born in Mexico for hypothyroid, for adrenal, for metabolic. You don't have that safety net.


Gong, Alice K 1:01:35
Mm-hmm.
No, you don't. Yeah, unfortunately that that is difficult to do. But if you get a baby that someone has that move, you can send that screen in, OK.

Jane Lynch 1:01:58
OK, that's great to know. I I mean, I think we've done it, but and where do you get the card? You just what would be the easiest way to make it happen?


Gong, Alice K 1:02:06
The.
The hospital has them. I mean if you send them to the lab, they have them the in private practice you have to kind of buy those kids or you can say you have a Medicaid patient and then and get some, but.

Jane Lynch 1:02:11
OK.
Oh, OK, that's easy.


Gong, Alice K 1:02:25

It's very nice. I mean the hospital. I see people going to the lab to get them drawn for their second screen. So they have to, they have the kids. You just have to provide all the information for it. OK and chat.

Jane Lynch 1:02:36
Perfect. Thanks.

Gong, Alice K 1:02:40
The best way is a website, OK. And so certain people have access to that website. If you're a provider, you can get access. But for us like in the we have people that work in in the NICU that have access. So they they get the information, they can go in and get.
Now the hospital, our hospital actually has it where we have a direct connection to the lab. So when we send a specimen from our hospital, when the results come back, it will show up in the medical record.
Which is amazing. You know, I have to go tracking it down.

Ranch, Daniel 1:03:23
All right. Well, if there aren't any other questions, thank you again, Dr. Gong, for that fantastic presentation. Thank you everyone for attending Grand Rounds today. Please don't forget to complete your post Grand Rounds survey and provide feedback. That data helps our speakers and also our program as well. So otherwise.
Have a great day.

1:03:48
Thank you.

Calderon, Delia stopped transcription