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**Webinar: Understanding Newborn Screening for Rare Diseases**

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# Transcript

Rachel Jones/NPF 00:05
Hello, and welcome to the Evelyn Y. Davis Studios of the National Press Foundation in Washington DC. My name is Rachel Jones and I'm the director of journalism initiatives within NPF.

Rachel Jones/NPF 00:18
Thank you for joining us for the reporting on newborn screening and rare diseases webinar. Today's event is sponsored by PTC Therapeutics, a global biopharmaceutical company that delivers transformative therapies for people living with rare diseases. For 25 years, PTC Therapeutics has harnessed its scientific platforms to create new therapies that address the underlying causes of disease and deliver on their promise to support patients and families.

Rachel Jones/NPF 00:51
You can learn more about their work through their website at PTCBio.com.

Rachel Jones/NPF 00:58
February 28th is the annual Rare Disease Day, and for many families, the first encounter with these complex medical conditions occurs at birth. Just moments ago, the National Organization for Rare Disorders released its latest white paper entitled, Preserving Public Trust in the U.S. Newborn Screening System. The analysis explores the issues surrounding a procedure that begins shortly after birth each year for about four million American babies who get a heel prick that yields a drop of blood. That drop gets analyzed for illnesses like Phenylketonuria or PKU, cystic fibrosis, and sickle cell disease.

Rachel Jones/NPF 01:45
But in this era of technological advances, how can families be assured that newborn screening and the genetic information it yields will produce vital treatments that will benefit people living with rare disease?

Rachel Jones/NPF 02:12
To answer that question, we're joined by three experts who will help journalists understand the science and the real world impact of newborn screening. First, we're joined by Allison Herity. She's a senior policy analyst with the National Organization for Rare Disorders and a principal author of today's newborn screening study. As a rare disease patient herself, Herity's work as a researcher helps translate research and policy into real world solutions for families and patients. Next, Sarita Edwards is the CEO and president of The EWE Foundation and the host of the Being Rare podcast. The EWE Foundation was founded after Edwards' son Elijah was born with the rare disease Trisomy 18. And she'll help journalists understand the role that newborn screening played in her family's diagnostic odyssey. Our third panelist is veteran science journalist Bijal Trivedi, whose extensive reporting and writing background includes the book Breath from Salt, A Deadly Genetic Disease, A New Era in Science, and the Patients and Families Who Changed Medicine Forever. Her deep dive expertise into genetic research related to diseases like cystic fibrosis and sickle cell anemia provides crucial insights for journalists who want to add context to their reporting about rare diseases. You can read the full bios of our panelists on our website at nationalpress.org.

Rachel Jones/NPF 03:41
Thank you all for joining us today. Um, Allison, I'd like to start with you. Before you give us the details uh about the white paper that is released today, please start by giving the journalists a brief primer about the history of newborn screening and where the US stands today in terms of public health policies around that practice.

Bijal Trivedi/Science Journalist and Author 04:04
Sure, thank you Rachel, and uh thank you for the opportunity to speak about newborn screening today. Um, as Rachel said, my name is Allison Herity and I'm a senior policy analyst with the National Organization for Rare Disorders or NORD. Um, the policy portfolio that I cover at NORD focuses on policies that affect access to diagnostics, including newborn screening, genetic testing, as well as the intersection of the two. Uh NORD is a non-profit organization dedicated to helping individuals impacted by rare diseases, organizations that serve them. Along with our over 350 patient organization members, we work

to advance the identification, treatment, and cure of rare disorders through a wide range of programs including education, policy and advocacy, research, and patient support services. Um, many people with rare conditions face years of a difficult diagnostic odyssey before receiving a correct diagnosis. This can be a really difficult time for people who are impacted by rare conditions and their families, and in many case cases, these delayed diagnoses can result in mental and financial strain, suboptimal care, and irreversible health complications. I'm going to provide a bit of a general overview of newborn screening and discuss the current newborn screening policy landscape. What I hope that you'll take away from this conversation is that newborn screening is a vital lifesaving public health program that there are current challenges that pose a threat to the continued successes of the program, but that we have the opportunity to address these challenges and strengthen our newborn screening system for the future. At the outset of the conversation, I want to highlight a distinction that I feel is important to keep straight as a policy professional working in this space and for journalists who are reporting on these topics. And that's the distinction between newborn screening as we see it today and newborn sequencing. Newborn screening is a population-level public health program that predates the Human Genome Project and the genomic sequencing technology we have available today in 2025. It actually originated in the 1960s when a doctor named Robert Guthrie developed a blood test for a condition that Rachel mentioned earlier called Phenylketonuria or PKU. Babies that are born with PKU uh appear healthy at birth but are born with insufficient levels of an enzyme that's necessary to break down specific components of a protein. As a result, an amino acid called phenylalanine builds up in the body and causes permanent neurological damage. Prior to Dr. Guthrie developing this blood test for PKU, kids with PKU weren't diagnosed until after they started to show symptoms, at which point they had already developed permanent brain damage. The blood test allowed healthcare providers to detect the condition shortly after birth, giving them the opportunity to treat the condition earlier and avoid serious health complications.

Rachel Jones/NPF 08:41
I just I'll just jump in right here. I'm sorry Allison. I want to jump in right here and

Bijal Trivedi/Science Journalist and Author 09:21
Sure, yes. Um, I'm happy to dive into that. So, um talking a little bit about newbor the white paper that NORD has released today. So, the white paper that um we released focuses specifically on um the retention and secondary use of newborn screening dried blood spots leftover after screening is completed, which we refer to as residual dried blood spots. Um, these residual dried blood spots are a really important resource for newborn screening program operations as well as for public health and rare disease research. But controversy, unclear legal and ethical guardrails, and ambiguity around retention and secondary use policies may impact the continued use of residual dried blood spots and the success of lifesaving newborn screening programs more broadly. Um, the topic is complicated and requires a good deal of nuance, so I encourage everyone to check out the white paper and feel free to contact NORD if you have any questions or want to discuss the topic. And I can go into it a little bit more, but I also know that in the interest of time, um I'm not sure how deeply you want me to get into um the specifics of the paper. So, uh when you and I talked in our prep conversation, you talked a little bit about the, for example, the Early Check uh program in North Carolina uh and some of the the sort of contentious uh aspects of that. So can you talk a little bit about the white paper's findings in terms of what access to this genetic information means for communities and and what states are having to deal with in terms of the programs.

Sarita Edwards/E.WE Foundation 15:36
Yeah, absolutely. Thank you Rachel and thank you for the opportunity to be a part of this conversation. Um, back in 2016, my husband and I learned that we were pregnant with our fifth child and it was around 22 weeks pregnant that we learned of uh his rare condition, Trisomy 18 or Edwards Syndrome. Um, along with the diagnosis, we were told that he would die in utero, during delivery, or shortly after birth. Um, of course, receiving a diagnosis with a high mortality rate, um only 5 to 10% of babies who are born or diagnosed with this condition will live past their first birthday. And so um immediately my prenatal care changed. Um, our son was born alive in March of 2017, and um for us we were actually not given newborn screening because of the diagnosis. We were told that he looked consistent with everything that we had been told prenatally and um and that he was not worth the medical resources. It are actually the words that that were given to us. Um, we were sent home post delivery in hospice care where we stayed for seven months and it was after uh those seven months um we had a decision to make. We can continue forward with hospice or uh we can begin to pursue life sustaining measures and the conflict really was, I had already started, you know, care coordinating him myself, reaching out to doctors and trying to find out more about his diagnosis and and and so to me the option was stop doing that or continuing in hospice care and so we elected to uh, you know, stop hospice and move forward with trying to learn more about his condition. Um, so for us, you know, our we we we were not on a diagnostic odyssey like a lot of families in the rare disease space. Um, we did get our diagnosis in utero, um but because of that diagnosis, um we were not afforded newborn screening and um

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Rachel Jones/NPF 17:47
If I'm remembering correctly, uh your other children had undergone the process and so for you this was sort of out of the norm. Tell us about the moment you found out that there was another baby with a similar condition who actually received newborn screening. Yeah, we actually I learned of another family um in my home state um who had the exact same diagnosis that that my son has. Um, she had a daughter, I had a son. Um, medically her daughter was um um more critical than my son. However, after a conversation with her myself, I learned that she did undergo newborn screening. She also received um early interventions. She was not sent home in hospice care. Um, she was a um a scientist. Um, my background professionally is in healthcare administration. Um, and so um, you know, and I have to also say that she was a white a white family. She was she was a white mom and you know, with a white child and I do believe that race contributed to the disparity in care um or the difference in care. Um, and so after a conversation with her, I was just very shocked that we delivered at the exact same hospital one year apart from each other. Um, she was actually born her child was born before my son. Um, and so I was very shocked to meet her and hear of her experience and my son was born a year later and received what we received and so yeah, that was that was very very appalling to me.

Rachel Jones/NPF 19:32
I'm going to want to ask you talk about your insights about what having access to this information can mean for communities, but at this point I want to pivot to Bijal. Uh Bijal, your expertise and your uh work in reporting and writing about genetic illnesses is broadly known and and I know you've just gotten back actually from a trip to Nigeria where you've done some deep

Rachel Jones/NPF 20:00
reporting on sickle cell anemia, but talk about your work in reporting on the book Breath from Salt and what you learned about genetic testing and the role of early screening.

Bijal Trivedi/Science Journalist and Author 20:13
Um, when uh I started reporting for um my book Breath from Salt, which was all about the the history of developing cures and treatments for cystic fibrosis.

Bijal Trivedi/Science Journalist and Author 20:25
Um, I learned that back in 1976, um this guy named Phil Farrell, who was um a doctor, a pulmonologist at the University of Wisconsin,

Bijal Trivedi/Science Journalist and Author 20:38
he learned that the only way to really get a hold of cystic fibrosis and control how the the disease evolved um on a population scale was newborn screening.

Bijal Trivedi/Science Journalist and Author 20:51
Um, but because it wasn't an established practice. I mean it was established for um PKU in 1963,

Bijal Trivedi/Science Journalist and Author 21:04
but for most other diseases there was no basis for doing this. And because it's an expensive thing, um or it was back then, you know, he needed a rationale.

Bijal Trivedi/Science Journalist and Author 21:18
So he did um a randomized clinical trial um to show the impact of newborn screening on children with CF. And this is one of the most involved clinical trials that has been done for newborn screening and he

Bijal Trivedi/Science Journalist and Author 21:33
screened in the state of Wisconsin more than 650,000 children over the span of nine years. And he showed that those who were given a newborn screening test and the parents told that their child had cystic fibrosis,

Bijal Trivedi/Science Journalist and Author 21:43
those children had a much better survival rate. They grew normally, um they had fewer complications of lung infections, um and overall their health was better. They grew up stronger.

Bijal Trivedi/Science Journalist and Author 21:56
Uh those who did not receive um that diagnosis of newborn screening, um those children were sicker, they had lung

infections, they suffered from malnutrition, they were stunted and stunted refers to a case where you're not given nutrients in a particular stage of life and you really can't compensate later on for those growth abnormalities sort of mentally or physically.

Bijal Trivedi/Science Journalist and Author 22:15
And um he proved that that newborn screening was essential um to helping treat cystic fibrosis and to help those children survive to a certain age and live a good life.

Bijal Trivedi/Science Journalist and Author 22:29
Um, now he discovered this, the study was published in 1997. Um by 2005, um only five states required a test for cystic fibrosis

Bijal Trivedi/Science Journalist and Author 22:44
and it wasn't until um 2009 that all 50 states tested for it. Um but I wanted to add to this that, you know, later reporting I did after the book,

Bijal Trivedi/Science Journalist and Author 23:00
um revealed to me that just because a state has newborn screening does not mean that every state tests for the same mutations. And this can lead to um enormous health disparities.

Bijal Trivedi/Science Journalist and Author 23:18
And one case that I covered um for a story I wrote for National Geographic um in 2023 um was a case um where the patient Terry White, um

Bijal Trivedi/Science Journalist and Author 23:33
friend me. Um Terry is African-American, he presented with the the symptoms of uh cystic fibrosis when he was born and throughout his childhood and early adulthood and people looked at him and said, well,

Bijal Trivedi/Science Journalist and Author 23:51
it would seem that you have cystic fibrosis but you're black so you don't. Um so there have been multiple cases where

Bijal Trivedi/Science Journalist and Author 24:02
doctors have looked at patients and said, oh because of your ethnicity we don't think you have this disorder. But

Bijal Trivedi/Science Journalist and Author 24:13
that is because um just because a disease has been identified in a particular ethnicity. For example, cystic fibrosis is most common in people of European descent, does not mean the disease doesn't occur in other ethnicities.

Bijal Trivedi/Science Journalist and Author 24:33
And with newborn screening, even though all 50 states can screen for cystic fibrosis, it doesn't mean they're screening a wide range of mutations. So the mutations that cause a disease like cystic fibrosis, the most common one is called delta F 508, uh F508 delta.

Bijal Trivedi/Science Journalist and Author 24:54
And that it appears in Caucasian populations.

Bijal Trivedi/Science Journalist and Author 25:00
it's the most common mutation.

Bijal Trivedi/Science Journalist and Author 25:01
But this disease also occurs in Asians.It occurs in Egyptian people.It occurs in um quite a few people in the Middle East.And all those other countries and people of whose ancestors evolved there, um they carry different mutations.

Bijal Trivedi/Science Journalist and Author 25:21
Um so in some states they will test for just the Caucasian mutation and in other states they will test for a range of mutations.

Bijal Trivedi/Science Journalist and Author 25:32
So I just think it's important to realize that unless a state is using a newborn screening test that involves um an ethnically diverse range of mutations, you're going to miss people.And you're going to miss people of color and that's going to

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exacerbate health disparities among that population.

Bijal Trivedi/Science Journalist and Author 25:52
And that is such an important contextual piece uh particularly for journalists to understand and so uh

Rachel Jones/NPF 25:57
I'll get back to you to talk about your um latest project on sickle cell anemia but want to pivot back to Allison for a moment.

Rachel Jones/NPF 26:09
In my uh research uh around this topic, uh I believe I saw several figures but many of them were about 13,000 babies uh per year in the US are actually uh identified as having a rare disease and so I want to talk or you to talk about uh NORD's perspective and your perspective as a researcher

Allison Herrity/NORD 26:32
about this issue of uh continuity of of um practice in terms of which diseases are screened uh the financial aspect.Tell us give us some context about that piece of it.

Bijal Trivedi/Science Journalist and Author 26:46
Sure.So yeah, so um a large number of newborns are um identified to have a rare condition through newborn screening each year.

Bijal Trivedi/Science Journalist and Author 26:55
Um within newborn screening there is newborn blood spot screening which is typically what we are talking about when we're talking about being diagnosed with um rare conditions like through the heel prick and that's about 6,700 newborns are diagnosed through that every year.

Bijal Trivedi/Science Journalist and Author 27:10
When you add on congenital heart disease screening and early hearing screening is when you get to that 12,000 to 14,000 sorry um number.

Bijal Trivedi/Science Journalist and Author 27:20
But yes, kind of as you alluded to um and as I was mentioning earlier, these um newborn screening programs are run state to state and so um each

Bijal Trivedi/Science Journalist and Author 27:30
which state determines specifically what newborn screening conditions um are on their panel as well as what tests they're using to test for those conditions and so that um can create a situation where you have um a newborn potentially not being screened for a condition that had they been screen born in a different state that screens for that condition they um you know would have been screened.

Bijal Trivedi/Science Journalist and Author 27:36
And so NORD is supportive of and you know as I know many organizations in the space are of um screening for the conditions that are on the um Department of Health and Human Services recommended uniform screening panel for um kind of traditional newborn screening that we talk about right now but one of the things that is um something that we have to think about for the policy landscape in the future is as we have these emerging technologies um both when it comes to testing and being able to screen for more disorders cheaper and quicker as well as more treatments.

Bijal Trivedi/Science Journalist and Author 27:47
We have a really exciting cell and gene therapy pipeline right now. I think the figure is that we expect to see 60 cell and gene therapies coming

Bijal Trivedi/Science Journalist and Author 28:45
to market by 2030.

Bijal Trivedi/Science Journalist and Author 28:51
Um, you know, how can we support states uh in adding more conditions to their state newborn screening panel in a timely manner so that um, you know, newborns are able to access these critical treatments for the conditions identified.

Bijal Trivedi/Science Journalist and Author 29:41

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Sarita, in your work um with the podcasts as you talk to audiences, but also in your travels around the country um meeting families and patients with rare diseases.

Bijal Trivedi/Science Journalist and Author 29:52
What is your take on access to information about newborn screening, our communities really cognizant of of the um necessity for screening. What are you hearing?

Bijal Trivedi/Science Journalist and Author 30:00
says if newborn screening is being shared and what we learned was that 70% of our families had no knowledge of newborn screening. Um, they had no idea of what it was, even if they had other children before, um, they were they were not aware of the the details of newborn screening. They knew that their babies were receiving some level of tests after being born, they just did not know that it was newborn screening. Um, we also learned that because of the high mortality rate of the diagnosis,

Bijal Trivedi/Science Journalist and Author 30:33
5% of the families who completed our survey, 5% reported that they had received a diagnosis that uh newborn screening could could have identified had they been given newborn screening because we also wanted to know with your trisomy 18 diagnosis did you receive newborn screening. 5% of those families said we did not receive newborn screening but we did uncover a diagnosis later and um I think we had about uh 15% of families said that we received newborn screening and an additional diagnosis was discovered. So to us that's that's quantifiable. Um,

Bijal Trivedi/Science Journalist and Author 31:17
evidence that supports that there is an opportunity to better educate families but also better educate the healthcare space because what we're seeing is the numbers are telling us that the healthcare space needs to also um understand that regardless of a diagnosis, regardless of how critical it is, there is opportunity to uh complete newborn screening on these infants and still have an opportunity to to create a condition. One child we know for certain was diagnosed with hearing loss who later um had the opportunity to receive a care plan and is now currently hearing without the support of a hearing aid. We also know of a family who uh received uh the cystic fibrosis diagnosis by way of newborn screening, um, and they were able to better provide a care plan for that child because newborn screening identified it. Had she not received newborn screening they would have solely looked at that trisomy 18 diagnosis and unfortunately in the healthcare space trisomy 18 is considered incompatible with life. It does give leverage to a lot of doctors to refuse us care. Um, we have been denied healthcare. Um, and so our research is is strongly supporting that there is an opportunity for the healthcare space to um better educate families on newborn screening but we're also leveraging what we're finding to empower families so they know to ask these questions if it's not being presented to them.

Bijal Trivedi/Science Journalist and Author 32:52
Bijal from a a journalist's perspective obviously what those of us who cover health issues, one of the things that is often on our radar screen is the cost of uh treatments or screening or something that as journalists we can wrap our minds around fairly quickly. But in my prep conversation with you, you brought up what I thought was an interesting point which is that uh in some ways and may I may be paraphrasing it wrongly but um it can cost as much to to screen for fifty conditions as it does for just the one. So can you talk a little bit and give us a little bit of context about this issue of costs?

Bijal Trivedi/Science Journalist and Author 33:36
Sure. Um, what I was referring to then is for example in um Mississippi at the time I I wrote the story, um they screened for one mutation in the population um to detect cystic fibrosis. Um, they screen for the delta f508 mutation. Um, and as a result they missed 53.7% of African Americans who were suffering from cystic fibrosis. Now in other states they screened for 139 mutations in the CF gene and they detected 83.4% of African Americans who suffered from the disease. So just by screening a broader number of mutations you have the a much greater ability to detect the disease in other ethnicities and testing for one mutation in the CF gene does not cost any more than screening for 139 mutations. So that's that's where you can really increase the power of newborn screening if you if you test for multiple mutations in a range of ethnicities. Um, you know this is something that has to be ongoing and when

Bijal Trivedi/Science Journalist and Author 35:00
research insists on using a diverse selection of people for its research. That's critical to getting more information about how the disease presents in different populations and understanding which mutations cause the disease in which populations. So you screen for one CF mutation, why not screen for, you know, 200? We have the knowledge, we know those mutations, we

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get we have more and more information about which populations suffer, you know, are get the disease based on different mutations.

Bijal Trivedi/Science Journalist and Author 35:18
So it really makes no sense not to to screen for more.

Bijal Trivedi/Science Journalist and Author 35:41
Let's go back to you Alison for a minute because what I want you to sort of uh address is this issue of how newborn screening can eventually lead to treatments and better research and what it is that in putting together this white paper and in your expertise

Bijal Trivedi/Science Journalist and Author 35:46
uh help journalists understand the role to the path to better treatments.

Bijal Trivedi/Science Journalist and Author 36:07
Sure. So um, you know, one of the big things that um, you know, was a catalyst kind of for us to uh do a deeper dive into this newborn screening dried blood spot issue and um write this white paper is that the, you know, as I mentioned the newborn screening dried blood spots are a really valuable resource for public health research but also for rare disease research. These conditions obviously are rare conditions and so it can be difficult to find, you know, enough samples to um do certain research studies and especially when we're looking at um developing new screens for rare conditions specifically. There's a huge need for um, you know, access to blood spots, especially blood spots for those, you know, for newborns who've been identified to have these conditions. Um and when states destroy them really quickly, um one of the problems that you can have is that that newborn's blood spot who's been identified to have, you know, homocystinuria for example, um might have already been destroyed in a state that destroys uh blood spots very quickly before the child has ever had the chance to have confirmatory testing to, you know, follow up and see that they actually have the condition.

Bijal Trivedi/Science Journalist and Author 36:57
And so then you've lost access to that blood spot that could be used um, you know, to help improve screens for homocystinuria or to help improve um, you know, treatment efforts and things like that in the future. Um and you know, they can be used for other types of research as well both for, you know, rare conditions but also for things like environmental toxin exposure research and things like that that are very important um and you need these, you know, large population-wide um databases to conduct research on.

Bijal Trivedi/Science Journalist and Author 37:51
Sarita, um tell us a little bit about how Elijah is doing and also help us put into context uh what difference it might have made had he been screened early on in terms of his treatment uh plans and progress. Um well today Elijah is is eight years old. Um he is in second grade. He is in our public school special education program. He is doing very very well um completely defying the the odds that we were given. Um and um a lot of the information that we were told, you know, we truly believe that had we followed the plan of care that we were given for Elijah, we would not be celebrating his eighth birthday um in just a few short weeks. Um I think in terms of, you know, had we received newborn screening for him, I truly believe that um we would have uncovered um some underlying conditions, you know, just some comorbidities that go with his his trisomy 18 diagnosis. Um we would have been able to start some intervention plans a lot sooner than we did. Um thankfully, um that that delay, that denial did not hinder Elijah from reaching the milestone that he has. Um we just unfortunately did not, you know, have the opportunity to address those concerns sooner. Um a lot of those a lot of the issues that that newborn screening potentially could have uncovered for us, we didn't begin to explore until Elijah was close to a year old because that's when the conversations started. And so it was it was truly um deciding to not move forward with another round of hospice care for him that

Bijal Trivedi/Science Journalist and Author 40:00
that that that really started to change the entire outcome and the trajectory of trajectory of his care um and so so yeah we just truly believe that had had he received newborn screening we we we believe um we would have been able to take on some of those challenges a lot sooner than we did.

Bijal Trivedi/Science Journalist and Author 40:22
Bridgette, uh let's talk about your reporting in Nigeria and sort of the global perspective uh of uh what having a broad base of genetic information uh can mean in terms of of developing treatments for rare disease. You focused on sickle cell and your upcoming book will be about that. Is that correct?

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Bijal Trivedi/Science Journalist and Author 40:43
That's right. That's right. Um so I had um two pretty extraordinary reporting trips, one to India and one to Nigeria and

Bijal Trivedi/Science Journalist and Author 40:53
the the pictures I got of newborn screening in both cases were were really interesting and quite different. Um

Bijal Trivedi/Science Journalist and Author 41:04
I think the biggest problem when you're talking about uh places that are resource poor is that people are afraid of this knowledge. People are afraid of hearing the result of a newborn screen or a genetic screen in general because there's so much stigmatization of these diseases um both in India and in Nigeria and

Bijal Trivedi/Science Journalist and Author 41:28
what was really surprising in Nigeria was that they had um they had a beautiful system for presenting this genetic information to mothers and then having a genetic counseling session depending on the genotype of their child. So if the child had sickle cell, they would be signed up for a different genetic counseling session than somebody who was a carrier of the disease. And I sat in on these genetic counseling sessions um pardon me, each of which had about 30 women and it was amazing because they were told how the disease is transmitted, which is not known in many cases um because it's not part of their biology program in say high school. Um so the nurses are um tasked with explaining how the disease is transmitted that it's not a curse that it is not um a death wish which is the idea that many of the women had and that you know with um

Bijal Trivedi/Science Journalist and Author 42:07
with the proper care these children can thrive. Um and I think the you know when the newborn screening goes hand in hand with that genetic counseling the the results were extraordinary. Um the nurses introduced me to mothers who were absolutely devastated by that diagnosis several years ago and now they were bringing their children in the children were healthy, they were thriving and it was it was really an uplifting story um and to see how these children could get the attention they need because the earlier your um

Bijal Trivedi/Science Journalist and Author 42:56
diagnosed with sickle cell the earlier you can be screened for things like strokes and that's one of the most debilitating um consequences of having sickle cell is strokes from age two to about age seven. Uh so these children can be monitored, their nutrition can be um improved and they can be given um regular penicillin to decrease the number of infections. So there was you know a very positive example of what newborn screening can do. Um in other places it's it's just much harder to educate both the physicians and the population and when it comes to diseases like sickle cell you have to raise awareness on a national level and that's where they are in India.

Bijal Trivedi/Science Journalist and Author 43:40
Allison, uh I really as we wind this conversation down I really hope that you can help the journalists sort of understand and uh the tension between concerns about what's being done with this information versus potential to to help and provide

Bijal Trivedi/Science Journalist and Author 44:41
right solutions.

Bijal Trivedi/Science Journalist and Author 44:43
So can we we sort of get back to this issue of communities and states being worried about how these blood spots are going to be used versus the enormous possibilities of yielding a cure.

Bijal Trivedi/Science Journalist and Author 44:48
What how can journalists try to communicate that?

Bijal Trivedi/Science Journalist and Author 44:49
Sure, yeah.

Bijal Trivedi/Science Journalist and Author 44:55
I mean I think there's definitely as you said kind of this tension between um the necessity to keep newborn screening dried blood spots for some period of time.

Bijal Trivedi/Science Journalist and Author 45:05
As I mentioned before, they're critical to the functioning of newborn screening programs and they're you wouldn't be able to

operate newborn screening programs 100% from contrived samples, for example.

Bijal Trivedi/Science Journalist and Author 45:11
You need actual newborn screening dried blood spots to continue doing that.

Bijal Trivedi/Science Journalist and Author 45:18
And you know, we want to continue benefiting from the research um aspects and benefits that dried blood spots provide.

Bijal Trivedi/Science Journalist and Author 45:21
But one of the really big points we want to drive home is that, you know, once you explain to people who are unfamiliar with newborn screening, which a lot of people are unfamiliar with newborn screening, but once you explain it to them and you say, you know, these are life-threatening potentially conditions that there's a treatment for, but you have to identify them early or, you know, by the time that symptoms present it, you know, is too late to prevent some of these um complications that are happening.

Bijal Trivedi/Science Journalist and Author 45:23
People are supportive of the program, but we need to go at another level and explain, you know, because if people have the question of, okay, like I understand that, I'm supportive of that, but why do you need to keep the blood spots?

Bijal Trivedi/Science Journalist and Author 45:48
And we need to do a much better job, I think, um both, you know, in the uh, you know, patient advocacy space and just in general of drawing that direct line between, you know, we need to keep these dried blood spots in order for us to benefit from the life-saving aspects of newborn screening.

Bijal Trivedi/Science Journalist and Author 46:00
Recognizing that, I do think and something that we talk about in our white paper is that um, you know, newborn screening programs could be much more transparent about the fact that they are keeping these samples um for periods of time and what they're being used for.

Bijal Trivedi/Science Journalist and Author 46:01
Um, one of the studies that we looked at and that we highlight in the paper um showed

Bijal Trivedi/Science Journalist and Author 47:02
NORD that mothers that were um given education about both newborn screening and about dried blood spot um retention and secondary use were more supportive of newborn screening overall as well as of dried blood spot retention.

Bijal Trivedi/Science Journalist and Author 47:04
And so, you know, making sure that um, you know, there are easy steps that we can take to be a bit more transparent about um, you know, what that these are being uh kept and, you know, why they're being kept and what kind of, you know, research and things they're contributing to.

Bijal Trivedi/Science Journalist and Author 47:22
Um to help, you know, make it more known that um the importance of keeping these samples.

Bijal Trivedi/Science Journalist and Author 47:51
Bijal, are we possibly in an era of um pretty significant opportunity when it comes to harnessing information, uh genetic information that newborn screening can yield uh and furthering a pathway to treatments, better treatments.

Bijal Trivedi/Science Journalist and Author 47:53
Uh is there an opportunity that we have or a threat of it being sort of choked off because of fears and concerns about, you know, access to information.

Bijal Trivedi/Science Journalist and Author 48:14
Um as uh as an eternal optimist, I I have to say that I think um there's an enormous opportunity to improve health of all children um using newborn screening.

Bijal Trivedi/Science Journalist and Author 48:52
And that's because, you know, as Alison and Sarita have have noted that, you know, the earlier you diagnose uh an illness, the easier it is to treat. And with newborn screening these are these are conditions where early diagnosis is key to the health

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of that child beginning on day one or day two um of their lives and the the it it's something that will affect the health um the mental health and the physical health of that child forever.

Bijal Trivedi/Science Journalist and Author 49:23
I think you can't just

Bijal Trivedi/Science Journalist and Author 49:24
reserve newborn screening for one subset of the population.

Bijal Trivedi/Science Journalist and Author 49:30
You have to do this for everyone, every child of every ethnicity and not to get on a soapbox here,

Bijal Trivedi/Science Journalist and Author 49:37
but I'm getting on a soapbox here.

Bijal Trivedi/Science Journalist and Author 49:42
Um, you have to screen for mutations that occur in all people of all ethnicities.

Bijal Trivedi/Science Journalist and Author 49:48
Otherwise every child is not getting an equal chance at at good health and I think that's

Bijal Trivedi/Science Journalist and Author 49:53
so unethical, um,

Bijal Trivedi/Science Journalist and Author 49:58
you know, to distinguish between particular groups of people saying this group will screen for everything and they'll be in great health

Bijal Trivedi/Science Journalist and Author 50:03
and this group, well, you know, we'll screen for some mutations but not all of them.

Bijal Trivedi/Science Journalist and Author 50:09
So it's an opportunity I think because, you know, I believe that science should be used for good at improving health.

Bijal Trivedi/Science Journalist and Author 50:15
Um, and so that's my Pollyannish, not Pollyannish,

Bijal Trivedi/Science Journalist and Author 50:21
you know, um, view on it.

Bijal Trivedi/Science Journalist and Author 50:28
Well, we we appreciate it and it makes sense.

Bijal Trivedi/Science Journalist and Author 50:32
As we wind down, I think what I'd like to ask each of you to do is, you know, as I mentioned earlier, Rare Disease Day is February 28th.

Bijal Trivedi/Science Journalist and Author 50:39
And so if you had your magic wand or you had your wish list of the kinds of stories that you might like to see related to this topic,

Bijal Trivedi/Science Journalist and Author 50:46
um, and give journalists some tips on how to produce them.

Bijal Trivedi/Science Journalist and Author 50:52
Let's start with you, Sarita.

Bijal Trivedi/Science Journalist and Author 50:53
What um kinds of stories you like to see?

Sarita Edwards/E.WE Foundation 50:59

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You know, I would love to see a story that highlights um what a rare disease is.

Sarita Edwards/E.WE Foundation 51:06
You know, just start at the basics and um, you know, talk about what what is a rare disease, talk about um, you know, the community that you're in or the

Sarita Edwards/E.WE Foundation 51:13
and and when I say community, look at the entire state, you know, in in this state,

Sarita Edwards/E.WE Foundation 51:20
there are, you know, these many people um and and I'm I'm willing to bet that there are hundreds of thousands of people affected by a rare diagnosis right where you live.

Sarita Edwards/E.WE Foundation 51:32
Highlight those stories and just let raise awareness and let people know that in your own community, your neighbor could be someone dealing with a rare diagnosis and you just

Sarita Edwards/E.WE Foundation 51:39
don't know, you know, you you may notice that their child is always sick, but you don't know that it's a rare diagnosis.

Sarita Edwards/E.WE Foundation 51:46
Um, talk about the impact of rare diagnosis and what that means for families, you know, did a lot of families transition from two incomes to one.

Sarita Edwards/E.WE Foundation 51:53
Talk about the financial impacts.

Sarita Edwards/E.WE Foundation 52:00
I think that there's just a lot of opportunity to highlight just at the foundation level, highlight what rare diagnoses are, what they mean for your community and other and

Sarita Edwards/E.WE Foundation 52:07
ways that that, you know, you can be involved and and and I think it it would be really helpful too for journalists to reach out to patient advocacy organizations.

Sarita Edwards/E.WE Foundation 52:16
See see what what's happening in the community, see what stakeholders are invested.

Sarita Edwards/E.WE Foundation 52:24
Look at at the legislative landscape in in your state and see what's happening in terms of rediagnosis.

Sarita Edwards/E.WE Foundation 52:32
Um, um, look into do you have a center of excellence, do you have of a rare disease advisory council.

Bijal Trivedi/Science Journalist and Author 52:40
You know, those are all the things that I would love to see when I turn the news on to just highlight the importance of letting people know that it it may be a rare diagnosis, it may be a rare topic, but it really truly affects and impacts so many people.

Sarita Edwards/E.WE Foundation 52:47
I don't even understand why we don't talk about it more than we do already.

Rachel Jones/NPF 52:52
Actually a very good point.

Rachel Jones/NPF 52:56
We're starting to get some questions in right at the end.

Rachel Jones/NPF 53:03
So as I pivot to you Allison, uh one of the questions are what are there any new conditions being considered for newborn blood spot test testing

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Rachel Jones/NPF 53:10
and how can journalists sort of uh get at that aspect of the story?

Allison Herrity/NORD 53:22
Yeah, so I know that um Duchenne muscular dystrophy is currently going through the RUSP nomination process attempting to get on to the the RUSP being the recommended uniform screening panel, which is um a panel that the federal government uh the Secretary of HHS um

Allison Herrity/NORD 53:30
maintains with help from the advisory committee on heritable disorders in newborns and children.

Allison Herrity/NORD 53:40
Um, and you know that's another aspect of the newborn screening policy landscape is the the kind of the challenges that the RUSP nomination process poses for um often small patient advocacy or

Allison Herrity/NORD 54:05
organizations that are um, you know, the people who are putting together these nomination packages and driving them forward.

Allison Herrity/NORD 54:10
It's um a lot of the organizations that are NORD member organizations for example are an executive director and that is the staff of the organization and um, you know, that can be a big burden um

Allison Herrity/NORD 54:22
for these organizations to pull together and I I know that that's something that has been talked about a lot um in at least the rare disease policy space as it relates to newborn screening um and the RUSP.

Allison Herrity/NORD 54:43
And then I imagine I think put was put in the chat as well that metachromatic leukodystrophy or MLD will likely pursue RUSP nomination soon if they're not already doing so.

Allison Herrity/NORD 55:03
Um but those are the two that come to

Allison Herrity/NORD 55:05
the top of my head. It sounds like journalists might uh benefit from trying to find out which which conditions in their

Allison Herrity/NORD 55:12
cities, states, communities are screened and perhaps doing a deeper dive into that.

Rachel Jones/NPF 55:15
But Bijal, let's wind up with you.

Bijal Trivedi/Science Journalist and Author 55:17
Uh one of the things that I found in my years of reporting and working with journalists is there can be a bit of uh intimidation

Bijal Trivedi/Science Journalist and Author 55:27
about covering these kinds of topics. Uh health and science can seem a little bit weighty for them, but what advice would you give to journalists about perhaps taking a a

Bijal Trivedi/Science Journalist and Author 55:40
crack at reporting on this topic in their communities? Um

Bijal Trivedi/Science Journalist and Author 55:45
I am always moved by stories that focus on a family. For example, Sarita's family and I'm just I'm still boggled by your your story of the other family that was given

Bijal Trivedi/Science Journalist and Author 56:02
a different diagnosis. Um so I I love stories that show those contrasts because I think it's one thing to show the numbers um and another thing to show the implications of that of that decision not to give you newborn screening to

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Bijal Trivedi/Science Journalist and Author 56:28
to tell you to, you know, rely on hospice and sort of discard your child. Um that is incredibly powerful. Um so those sort of anecdotes while they are hard to get, I think are really worth the time because

Bijal Trivedi/Science Journalist and Author 56:43
you know, your story's gonna stick with me forever. I'll probably reach out to you later. Um but you know, if you can ground those those statistics in a story like Sarita's then I I just feel like those stories are the ones that can can change policy, can change minds and and that's really important.

Bijal Trivedi/Science Journalist and Author 56:55
I also think that looking into because newborn screening is so variable from state to state. I mean Mississippi state, you know, screens for I think it's 64 diseases, California screens for about 83.

Bijal Trivedi/Science Journalist and Author 57:12
You know, what's the difference there? Why? And who who, you know, what sort of medical care are you getting or not getting in Mississippi as a result of those newborn screening discrepancies, um state to state discrepancies.

Bijal Trivedi/Science Journalist and Author 57:25
So I think that's also a really important area to poke and one that can have, you know, if you succeed in writing a story on those, it's one that can translate to a lot of impact.

Rachel Jones/NPF 57:43
And on that very powerful note, I'd like to take this opportunity to thank our three

Rachel Jones/NPF 57:45
guests today, Allison Herity of NORD, Sweta Edwards of the Being Rare podcast and of the EWI Foundation and veteran science journalist and author Bijal Trivedi.

Rachel Jones/NPF 58:02
Thank you so much for joining the National Press Foundation's uh reporting on rare disease and newborn screening webinar.

Rachel Jones/NPF 58:11
I'd also like to take this opportunity to thank PTC Therapeutics for their support of our efforts to make good journalists better.

Rachel Jones/NPF 58:21
And to those watching today, we will be posting this video on our website at nationalpress.org and on our YouTube channel and we will also post a link to the NORD white paper that was just released today.

Rachel Jones/NPF 58:30
So thank you all for watching and please look out for our future NPFWebinars. Take care. Thank you. Thank you.

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