COMMONWEALTH OF PENNSYLVANIA
HOUSE OF REPRESENTATIVES

HUMAN SERVICES COMMITTEE
PUBLIC HEARING

STATE CAPITOL
HARRISBURG, PA

IRVIS OFFICE BUILDING
ROOM G-50

FRIDAY, APRIL 26, 2017
9:00 A.M.

PRESENTATION ON
IMPLEMENTATION AND FUNDING OF ACT 148 OF 2014,
PENNSYLVANIA'S NEWBORN SCREENING PROGRAM

BEFORE:
HONORABLE GENE DIGIROLAMO, MAJORITY CHAIRMAN
HONORABLE ALEXANDER CHARLTON
HONORABLE BUD COOK
HONORABLE MICHAEL CORR
HONORABLE CRIS DUSH
HONORABLE JONATHAN FRITZ
HONORABLE RICH IRVIN
HONORABLE AARON KAUFER
HONORABLE NICK MICCARELLI
HONORABLE TOM MURT
HONORABLE ERIC NELSON
HONORABLE TOM QUIGLEY
HONORABLE ERIC ROE
HONORABLE JUSTIN SIMMONS
HONORABLE TARAH TOOHIL
HONORABLE JUDY WARD
HONORABLE ANGEL CRUZ, DEMOCRATIC CHAIRMAN
HONORABLE JASON DAWKINS
HONORABLE MICHAEL DRISCOLL
HONORABLE STEPHEN KINSEY
HONORABLE MAUREEN MADDEN
HONORABLE JOANNA MCCLINTON

BEFORE (Cont'd):

HONORABLE DANIEL MILLER
HONORABLE MARK ROZZI
HONORABLE MIKE SCHLOSSBERG
HONORABLE BRIAN SIMS
HONORABLE PERRY WARREN

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I N D E X

TESTIFIERS

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SUBMITTED WRITTEN TESTIMONY

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(See submitted written testimony and handouts online.)

P R O C E E D I N G S

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MAJORITY CHAIRMAN DIGIROLAMO: Morning, everyone. I'd like to call this hearing and possibly voting meeting of the Human Services Committee to order and ask everyone if they would stand for a Pledge of Allegiance.

(The Pledge of Allegiance was recited.)

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Thank you. First, I'm going to ask Pam to take the roll.

(Roll was taken.)

MAJORITY CHAIRMAN DIGIROLAMO: Okay. A quorum being present, just set up some ground rules. It was my intention to take a vote on a bill first thing this morning. Some of you might be aware of the issue, the Consolidation Plan of the Governor, but there are some Members that are not present yet. I'm hoping to get them here so at some point during the hearing we might just stop and take the voting meeting.

But for right now, I think with everybody here, we can start with testimony. And I might ask Representative Cruz, my Democratic Chairman that asked for this hearing to be called, so if you want to open it up for a few comments.

DEMOCRATIC CHAIRMAN CRUZ: Thank you, Gene.

Thank you to everyone for coming in and putting your testimony that's so crucial and important facing especially the Krabbe disease.

I'm going to turn it over to my Legislative Assistant and Director to give us a brief explanation before we go into the testimony, and then we'll just go to the testimony. Thank you.

MS. MCCAHAN: Representative Cruz just wanted me to kind of run down how we got to this point. He had a piece of legislation that became law in 2014 that would expand some of the newborn screens we do in Pennsylvania under our Newborn Screening Program. That hasn't been completely implemented the way that his law was written, and so we're here just to get answers on that.

And he also wanted to bring your attention to a piece of legislation that he has introduced, House Bill 1081. That legislation would actually further modernize the Newborn Screening Program in Pennsylvania and create a funding mechanism for the program.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Okay. Yes, we're going to start. Our first testifier is not here yet, so we're going to start with Lesa Brackbill, who is the advocate and mother of Tori Brackbill. Welcome. Good to have you here.

MS. BRACKBILL: Good morning and thank you.

This is my daughter Tori and this is why I'm here today. Thank you for the opportunity to speak on behalf of H.B. 1081 and to tell you our story.

My name is Lesa Brackbill, and I live in Hershey with my husband, Brennan. We have a personal connection to this bill and to Act 148 of 2014, and I'd like to briefly share that with you.

Our daughter Victoria was born on July 30, 2014, in Harrisburg and she was perfect. She was healthy and smart and she met all milestones until five months of age. All of a sudden, she became increasingly irritable, stopped eating, lost weight, and no longer smiled, laughed, or played.

Six weeks after symptoms began, and just two weeks after we took her to Milton S. Hershey Medical Center for a CAT scan, the doctors at Hershey diagnosed our baby girl with Krabbe leukodystrophy and told us that it was terminal. On February 13, 2015, our lives changed forever. To make matters worse, they informed us that because it was genetic, we shouldn't try to have more children naturally.

Tori was the third child in three years to be diagnosed with Krabbe at Hershey Medical Center, and we are so thankful that they are currently the only hospital in the entire State screening each and every baby for Krabbe automatically.

We were told that there was nothing they could do to treat the disease and that she would pass away before age two. And they were right; there was nothing we could have done at that point. However, we quickly learned that if they had screened Tori at birth for Krabbe, they could have treated it, and that the treatment, though complicated, has a very high success rate. I'm not sure which news was worse: that our daughter was dying or that they could have done something about it just six months earlier.

We then learned that in October 2014 a bill had been signed into law making Krabbe one of the mandatory newborn screenings but that it hadn't been implemented yet. And so our journey began of doing everything we could to help Hannah's Law be used as it was intended.

I have attended the meetings of the Department of Health's Newborn Screening Advisory Panel for the past year and have been encouraged by the progress that has been made in educating the panel about the treatment for Krabbe and its effectiveness. Though the panel did not see things our way initially, we're all basically on the same page now, and for that, Brennan and I are grateful.

Through these meetings, I have learned a great deal about Pennsylvania's Newborn Screening Program, and I still cannot believe that we are nearly last in the Nation for the number of mandatory tests that are done in our Newborn Screening Program. You are likely aware of this, but each hospital in Pennsylvania chooses which diseases for which they will screen because currently they absorb the cost for all but the mandatory panel of nine diseases. The remaining 27 on the supplemental panel are not mandatory. This means that in our State, your ZIP code literally determines your life or death if you are born with a disease like Krabbe. This simply isn't right.

This bill would change that by merging the two panels so that all diseases are equally screened and every baby has a chance at life. However, there's a significant hurdle which holds back Hannah's Law and endangers the lives of children every day, and that's money. There's not enough money, we're told. This bill would change that as well. By changing to a fee-based system, as most States already use, the State would have the necessary funds to ensure that all babies are screened equally and for as many diseases as possible.

Money shouldn't be the determining factor in doing everything that we can to save the lives of babies who are born with genetic diseases. Whether it would be paid for by the parents or by insurance -- and insurance currently pays for much more frivolous things during the labor and delivery process -- it is a much-needed change. As a parent, I'd rather pay for newborn screening than for a funeral, and that's what we had to do.

Our daughter passed away last year, on March 27th at 20 months of age. She never walked or talked, she never smiled again, and she was robbed of the opportunity to live a full life, all because Krabbe wasn't caught quickly enough. And we as parents were robbed of the opportunity to do everything that we could to try to save our daughter's life.

Making H.B. 1081 law will help ensure that babies are given the best possible chance of life no matter which of the 36 diseases on the Pennsylvania panels are in their DNA. It was too late for Tori, but it doesn't have to be too late for other Pennsylvania babies who will be born with this or one of the other 35 horrific diseases.

Thank you in advance for making a difference in the lives of families in Pennsylvania.

MAJORITY CHAIRMAN DIGIROLAMO: Lesa, thank you very, very much for that very compelling testimony.

And, Chairman Cruz, I thought maybe we'd go through the agenda first --

DEMOCRATIC CHAIRMAN CRUZ: Okay.

MAJORITY CHAIRMAN DIGIROLAMO: -- before maybe we open it up for some questions --

DEMOCRATIC CHAIRMAN CRUZ: Sure.

MAJORITY CHAIRMAN DIGIROLAMO: -- and answers if that would be okay with you.

DEMOCRATIC CHAIRMAN CRUZ: Yes, sir.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Next on the agenda we have Dr. Michael Yudell, who's the Associate Professor and Chair of Community Health and Prevention, Drexel University, and Vice Chair of the Pennsylvania Newborn Screening and Follow-Up Technical Advisory Board.

Mike, how are you doing?

DR. YUDELL: Good, thank you.

MAJORITY CHAIRMAN DIGIROLAMO: Good. Welcome.

DR. YUDELL: Thank you. Representative DiGirolamo, Representative Cruz, thank you so much for having me this morning. It's a great honor and pleasure for me to be here today as the Vice Chair of the Pennsylvania Newborn Screening Technical Advisory Board.

A little bit about me, as you said, I'm Chair and Associate Professor of the Department of Community Health and Prevention at the Dornsife School of Public Health at Drexel. I'm a trained ethicist and historian, having received my Ph.D. and master's in public health from Columbia University. As such, I am dedicated to the just and ethical treatment of babies and families who are screened at birth in the Pennsylvania Newborn Screening Program, a critical public health program that saves and improves the lives of babies and their families each year.

As my colleague Dr. Jerry Vockley, the Chair of the Technical Advisory Board, as stated in his submitted written testimony, the TAB was constituted into law in 1980 to provide advice regarding newborn screening to the Secretary of Health. We do not make or enforce policy. Instead, the committee's collective expertise, drawn from a distinguished group of clinicians and scientists, makes recommendations to the Secretary based on our understanding and analysis of the best science. We make decisions based on specific criteria that include whether the testing has a significant impact on babies, whether a sensitive and specific screening test is available, whether early treatments of the disease affect its outcomes, and whether the resources exist to pay for screening follow-up and support of infants who test positive.

Now, as an ethicist, my role on the committee is to help the committee think through some of the ethical challenges of newborn screening. One recent example, we've been discussing Pompe disease on the committee. Pompe disease, as shown from the screening that's been in effect over the last year, has shown a number of late-onset cases meaning that we're testing for a condition that can appear later in childhood or even in adulthood. This has implications for the program. Does that force us to consider the nature of how we should be consenting parents who enter -- you know, whose children have their blood spots collected for the program.

The nature of the program's current opt-out consent is based on identifying diseases that require treatment at or shortly after birth and should the calculus change if we're telling parents that their child's condition might not appear until adolescence or later. Will those children face stigma or discrimination and might a family seek to take action contrary to the child's interests because of a late-onset diagnosis?

And of course the Newborn Screening Program doesn't bear responsibility of addressing all of these questions and concerns, but as testing technology advances in the future, and it surely will, Newborn Screening Programs around the country, including ours, will have to evolve and adapt to be able to provide resources to parents who will need to answer such questions.

To do this, it is essential for the program to have adequate resources, to be able to make recommendations to add and remove tests from the screening panel, to keep up with the latest science, and B) ensure that parents are educated on the nature of the program and its potential impact on babies.

The TAB has also recommended expanding the length of time blood spots are stored on filter paper by the State. This would allow us to use those blood spots for quality control, to demonstrate the utility of blood spots for other potential program efforts, to retrospectively test blood spots when a condition is added to a mandated test, and it could be used as part of research to develop novel screening markers for new tests. The program is currently unable to do this because of funding concerns.

For the program to be able to successfully educate the public about newborn screening, for the program to rapidly adapt to changing technological terrain and the expansion of testing, and for the program to have successful quality control and scientific reliability, the program must sit on sound financial footing.

That is why -- and I speak for both Dr. Vockley and myself -- that we are excited about House Bill 1081, which seeks to institute a newborn screening fee. Such a fee would go a long way towards providing a stable source of revenue to fund the program and its projects, particularly as there are increased mandates for screening and the program is forced to adapt to our rapidly changing technological age.

Finally, we appreciate the language in the bill deferring to the expertise of the TAB in adding and subtracting diseases from mandated screens. However, our committee is concerned about the addition of specific diseases to the list of mandated screens, an approach that would circumvent the TAB and could set a precedent that could have a significant impact on the program.

This approach, while driven by deep compassion for families, can contradict the best clinical evidence. The lysosomal storage disease Krabbe disease is among several new diseases currently listed as mandated tests in House Bill 1081. I would echo Dr. Vockley's statement from his written testimony that there is still insufficient medical evidence to yet change the TAB's previous recommendation on this and several other new diseases in the bill. We look forward to the day when successful interventions justify adding these conditions to the mandated tests. Thus, we advocate continuing to follow the recommendations of the RUSP and the best medical evidence as we make our recommendations to the Secretary.

Thank you for your time, and I'm happy to answer any questions.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Thank you. Would you stick around in case we have some questions afterwards?

DR. YUDELL: Absolutely.

MAJORITY CHAIRMAN DIGIROLAMO: Next, and our last testifier of the panel, Dr. Karen Murphy, who's the Pennsylvania Secretary of Health; and Dr. Rachel Levine, who is Pennsylvania's Physician General.

Welcome to the both of you.

SECRETARY MURPHY: Thank you very much, Mr. Chairman. It's a pleasure to be here, Chairman Cruz. I applaud your efforts for this hearing and also the efforts to protect newborns in the Commonwealth.

I can assure you that Ms. Brackbill's unimaginable loss is in our minds and hearts every day as we make all of our decisions.

So I'd like to first respond to what has happened over the last two years from a procedural standpoint, and then ask Dr. Levine to talk about the science that was behind our decisions.

So I rarely admit this, but between Dr. Levine and I, we have over 80 years of our life -- that shows our age so I don't like to bring that up much, but just to let you know what my background is is I'm a registered nurse, former CEO of a hospital that I worked at in my community for three decades, dedicated my career to health care and am very passionate about improving the health of Pennsylvanians. It's an honor to serve in this role.

So shortly after arriving in Harrisburg in 2015 and accepting the Cabinet position of Secretary of Health, this issue of the implementation of Representative Cruz's legislation came to me. And what accompanied that was a recommendation from the Technical Advisory Board, Dr. Vockley and also the entire committee. So when I researched it, what the role of the Technical Advisory Board, it's comprised of physicians, experts in the field to advise the Secretary on what should be done in regards to newborn screening.

The evidence that was support at that time, and as Ms. Brackbill stated, has evolved over the last year, but the evidence at that time was for us to select certain lysosomal screening tests and put them on the mandatory list. After listening and meeting with Dr. Vockley and the committee, we determined that we would follow the RUSP, which is again evidence-based, scientists that are saying what should be done for the health of newborns.

Again, I'm going to defer to Dr. Levine to talk about the science, but I can tell you that we support evidence-based medicine. We support anything that could improve the health of Pennsylvanians. I think when Dr. Levine talks about the science, you'll understand what our concerns are.

So I'm happy to follow up with any questions after Dr. Levine's testimony, and again, I thank you and thank you Representative Cruz for his work to modernize -- we should not be last in anything in health in Pennsylvania. Thank you.

DR. LEVINE: Thank you, Secretary Murphy.

So I'm very pleased to be here, Representative DiGirolamo, Representative Cruz, and the whole Committee. And in the last two-and-a-half years I've been very proud and honored to serve as the Physician General of the Commonwealth and working with Secretary Murphy and working with you all to advance the health of everyone in Pennsylvania.

So as we've been discussing, in terms of newborn screening, screening newborns for genetic endocrine and metabolic diseases is really a critical component of our public health mission, and newborn screening gives medical professionals the ability to detect and diagnose conditions that can affect the child's health and survival, and interventions can protect their health and prevent death and improve health outcomes.

The United States Health and Human Services convenes a panel called the Advisory Committee on Heritable Disorders in Newborns and Children, and they produce something called the Recommended Uniform Screening Panel, or the RUSP. So this is the Nation's finest scientists in this regard, and they produce a list of newborn screening. And every State has newborn screening programs even though they do it somewhat differently.

As has been discussed, Pennsylvania has the Newborn Screening and Follow-Up Technical Advisory Board, or the TAB, which is a group of Pennsylvania scientists and physicians which advise the Secretary of Health and the Department of Health on these issues.

In regards to lysosomal storage diseases, lysosomal storage diseases involve lack of an enzyme, so there's an accumulation of waste product in the cells that results in cell death and can result in severe medical complications and death. There are six lysosomal storage diseases that were added as part of newborn screening in your previous act, and they include two which are on the RUSP now so that the national board has recommended that they be screened for.

Now, what determines what the RUSP decides should be screened includes the quality of the testing, how sensitive and specific the testing is, how many false positives there'll be, how many false negatives there will be, and also critically, the availability of successful treatment that will affect the outcome of disease. And both those are very important points which I'll emphasize later.

So the RUSP in the last two years actually since we've been at the Department of Health have added two lysosomal storage diseases to the national testing panel, and that includes Pompe disease and something called MPS I or Hurler syndrome. The other four lysosomal storage diseases, including Krabbe, are not on the RUSP and not recommended by the national panel for testing, both because of the quality of the testing and in terms of the availability of successful treatment.

Now, New York, Missouri, and Kentucky have added Krabbe disease to their newborn screening, so there's three States in the country that have added Krabbe. There is no successful testing for the other three. There's no successful testing. There is no treatment at all for the other three lysosomal storage diseases.

For Krabbe, New York -- we're going to go over the New York experience -- began screening newborns for Krabbe in 2006. In an article that they published that I did reference in my testimony, although I don't know if the reference got there, but the article is called "Newborn Screening for Krabbe Disease in New York State: The First Eight Years' Experience." It's in a journal called *Genetics in Medicine*, and it was published in March 2016. And they outline the protocol in New York and their success.

And they had at the time of the article greater than 1.9 million babies that were screened. This is mandatory screening of all babies. They had 620 infants that had to have follow-up testing. Of that, 348 were referred to centers for even further testing. Five were diagnosed with Krabbe. Of that, one family decided not to do the treatment, which is a bone marrow transplantation in infancy in the newborn period, and that child passed. Two underwent transplantation and died from transplant-related complications. And two survived with moderate to severe handicaps. So this is the latest publication about the testing and treatment from New York State, which has the broadest experience for testing Krabbe.

So what we have been concerned about are those two aspects. Can the screening be done, which is sensitive and specific enough? So if you think about it, there were 620 infants whose families were notified that they might have a life-threatening and fatal condition. Of that, 348 went through that initial testing and had to be referred to a newborn screening, a genetic center in New York City for further testing. So you can imagine the anguish of those families. Five were diagnosed over that period of time. Four were treated. Two died in treatment, two had an outcome with moderate to severe handicaps. So we have concerns over the sensitivity and specificity of the testing, and we have concerns about the availability of a successful treatment.

So the researchers in the article question significantly about the utility from a public health point of view of doing this testing given those outcomes. There's a follow-up article, which is a commentary in *Genetics in Medicine*, and the conclusion of the follow-up article is that they do not recommend that States test for Krabbe at this time given the sensitivity and specificity of the testing and given the success of the treatment.

And that is the medical literature that has informed Dr. Vockley's comments, that has informed Dr. Yudell's comments, and informed the medical and scientific recommendations of the TAB.

Now, there are continued successes and advances in medicine all the time, and we are anxiously awaiting those advances. And so we had heard from Dr. Vockley that there might be some advances in the testing that they're just hearing about that might improve the sensitivity and specificity, so we had estimated that there might be in Pennsylvania 900 families a year who would have to have further screening, that there might be 2 or 300 that would have to actually go to a center to be further tested to diagnose a couple of infants, and the treatment was not very good. I mean, it wasn't very successful.

Now, there are advances in treatment, and I know that in one of the meetings that we had we spoke with a physician from Duke about advances that she had felt that she had made in terms of bone marrow transplantation, but that is not published. Dr. Vockley and I both did literature searches last week in preparation for this, and there is no new published literature from that physician or any other physician about what is different about their treatment, what is different about her protocol where she is saying that she's getting some better results.

So, you know, we live in a world where we have to rely upon evidence-based medicine, not verbal assurances that somebody's doing things better but what is in the medical literature, what is different about her potentially, that physician's protocol at Duke that she is getting better results? Could treatment centers in Pennsylvania then take that literature and maybe adjust theirs? But there's nothing published to recommend that.

As Dr. Murphy said, I mean, we've spent our live in the service of others in our medical careers and now at the Department of Health, and we absolutely do not want to see people suffer. And our hearts go out to the Brackbill family and the suffering that we had. But as of today, the science is not there to recommend universal screening for Krabbe. There's another article that I saw where genetic advisors, they don't recommend it. The scientists don't recommend that at this time because of the concerns about the testing sensitivity and specificity and the treatment.

However, things change. There might be an article coming out in several months which might change things. I mean, what we'd overall like to do is to follow the RUSP, is that we want to follow -- I mean, the idea of one uniform schedule as opposed to very interesting system we have where we have mandatory and then we have optional and supplemental. The idea of one schedule that follows the RUSP, that concept we support.

We don't have a specific opinion about your new bill because we're still looking at it with the Governor's office, but the concept of one uniform screening panel seems very progressive, but we want it to follow the science, and the latest science is determined by the RUSP and then our own TAB. Thank you.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Thank you both for your testimony. If you could stick around a couple minutes --

SECRETARY MURPHY: Sure.

MAJORITY CHAIRMAN DIGIROLAMO: -- we have one more testifier that's arrived, and then we're going to open it up for questions. Vicki? Our next testifier is Vicki Pizzulo, who is an advocate and mother of Hannah and someone who I know very well from Bucks County.

Vicki, good morning. Welcome.

MS. PIZZULO: Morning. Thank you.

MAJORITY CHAIRMAN DIGIROLAMO: And you can begin whenever you'd like.

MS. PIZZULO: I'm just going to tell my daughter's story.

Hannah Rose Ginion was born January 15th, 2013, weighing 8 pounds, 11 ounces. She was the biggest baby on the floor that day, so big that we thought healthy. Hannah had newborn screening done that was mandatory for the State.

We brought Hannah home to start our new exciting life with her, looking forward to hitting all our milestones. As the weeks went on, Hannah was progressing and meeting and exceeding her milestones. We were so blown away by how smart she was. She slept through the night and barely cried. We were so blessed in so many ways.

In May of 2013, Hannah started to slowly turn into a different baby. She was crying slightly more, I had trouble feeding her, and she was regressing her milestones. We addressed this to her pediatrician, and they said Hannah had acid reflux, and all babies meet milestones differently, not to worry.

Something still wasn't sitting right with us. We brought Hannah into the ER at Capital Health in Hopewell, New Jersey, due to Hannah not eating and just not being herself. You could clearly see something was wrong. They gave Hannah fluids and said Hannah has acid reflux. I asked the doctor, "How do you know that's what it is?" His words from a doctor, "I just watched her eat." I demanded tests to be done in the ER, and they told me no, I would have to come back and schedule tests.

We took Hannah home again. I made an appointment for an upper GI test and Children's Hospital in Philadelphia. The test came back that she had acid reflux. They said you usually don't see it, but with Hannah you did. This answer still did not sit right with me. Yes, I believe she had acid reflux, but I believe there was more.

A week or so later, we ended up back at Children's Hospital in Philadelphia, this time the ER. Hannah just stopped eating and she was crying all the time. Fast forward, we're being admitted for lots of testings. The neurologist had seen something in her CAT scan. We were going to do a test overnight to see if she had seizure activity. My baby had so many wires on her overnight.

The next morning at rounds, the neurologist came in and told me they were very worried for my baby. We were doing an MRI that afternoon. We went down for an MRI. My five-month-old baby was being put under. We were about to find out what is going on with our sweet baby girl.

June 6, 2013, at five o'clock p.m. two neurologists walk into Hannah's room. They sat us down and told us "Your daughter has something called Krabbe and it's fatal and she won't live past the age of two." We asked if there was a cure and they said it was too late for Hannah as it already progressed in her brain. If Hannah had been tested at birth, she could have received a lifesaving treatment, but PA does not test for Krabbe.

To give you a picture, Justin went into the corner crying, and I went over to Hannah's crib on my knees crying and begging to save my baby. She's just a baby. They said they couldn't, and they were crying as well.

Friday, June 7th, 2013, we had to figure out how to tell her sisters. This was a phone call I was dreading. How do you tell two little girls her sister is dying? I made that phone call, and as you can imagine, it wasn't good. All they kept saying was "I don't want my sister to die." I tried to calm them down the best I knew how at that point. I wasn't calm myself. We were losing our daughter. How is this even possible?

We spent a week at the hospital with more tests and trying to figure out Hannah's care. We were basically sent home with Tylenol and Valium and told to enjoy her. That answer was unacceptable to me. Children's Hospital wasn't familiar with the disease at that time.

I researched and I found a doctor who could help us better understand the disease and who could possibly give Hannah a lifesaving treatment. We rushed to Children's Hospital Pittsburgh, had more tests. Unfortunately, Hannah couldn't receive the transplant. The disease was progressing so fast from her last MRI. This is another reason why newborn screening is so important, but we were given medicine and learned how to give Hannah the best life she could get.

After returning home, we decided it's time to give this disease awareness. I was on Facebook and never heard of Krabbe ever. Also knowing that a test could be done at birth and how important it is to be done at birth, something needed to change. I needed to spread awareness. I needed to get a law passed to have mandatory Krabbe screening as a part of newborn screening.

We had a Facebook page created that was actually created while we were in the hospital to give updates to family. I called news stations and newspapers. I would tell anyone who would listen. This law, which would be known as Hannah's Law, needed to pass.

In August of 2013, just two months after Hannah's diagnosis, I received a call from Hunter's Hope. Angel Cruz was in touch with them to get Hannah's Law started.

He received a letter from one of Hannah's followers and was so touched by her story he wanted to help us. A few trips to Harrisburg and a few speeches, it wasn't easy. I spent countless hours trying to figure out how to get her story out there. Hannah was on every Philadelphia news station. Hannah had over 73,000 followers on her Facebook page. Countless people fell in love with our Hannah bear. And I did this all while caring for my sick child.

Finally, the day came, October 2014, the signing of Hannah's Law, an amazing day. This was right before we switched Governors. Again, I spoke with the Governor right beside me. I found the voice I never knew existed. You know why? Because we'd do anything for our children. I had to be Hannah's voice.

Let me tell you something from my speech that day. Most parents are excited to be celebrating their daughter's second birthday. I can't help but wonder if we'll be planning her funeral. Hannah passed away two months later after the signing at 23 months old.

I am disappointed that it's been three years and this law has not been implemented. Let me tell you, I hope anyone that is in charge of implementing this law doesn't ever have a family member born with Krabbe. What we go through as Krabbe parents is horrible, the countless hours we try to get them to breathe or their heartrate up or suctioning their mouth because they can't swallow. You have no idea. Every parent deserves the right to know if their baby has Krabbe at birth because if my Hannah bear, she would be here. If there is screening that must be done at birth in order to receive a lifesaving treatment, every baby should be tested.

My baby didn't even have a chance. Because of the Department of Health making these decisions for our baby, at the end of the day I'd rather have a false positive than a dead baby.

I will be forever grateful for Mr. Cruz and his team for continuing to fight for our babies and not stopping. He is a true hero in my book. He has no idea how much this means to me, me and every parent. Thank you.

MAJORITY CHAIRMAN DIGIROLAMO: Thank you, Vicki. You want to show us a picture of Hannah?

MS. PIZZULO: [inaudible].

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Thank you, Vicki.

I think we're going to open up for questions. Representative Cruz, you want to start off?

DEMOCRATIC CHAIRMAN CRUZ: Well, I just want to make a comment. You know, we've been working hard on this legislation. Governor Corbett signed this into law, and it still has not been implemented unless parents go and ask to get this test done. We have to change that. You know, we're not playing God here, but we're giving hope to families knowing that their children are at ease and not in so much pain.

So this is why I have the bill that will bring a mechanism and the funding of this Krabbe, so it is important, and I will continue the fight. I think it's important to newborns and families in Pennsylvania that they get treated exactly like everyone else and not because -- and Pennsylvania and Kentucky are the lowest newborn screening in the Nation, and we need to change that.

So I open it up for any of the Members who would like to --

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Representative Fritz.

REPRESENTATIVE FRITZ: Good morning, Mr. Chairman. Thank you. And to our two mothers, I just want to thank you for the bravery and strength and willingness to be here today and showing the pictures of your beautiful girls, thank you so much.

To our folks from the medical community, the dried blood spot screening, that method, will that produce the results for Krabbe? Can that method be used to detect Krabbe?

DR. YUDELL: Yes.

REPRESENTATIVE FRITZ: Absolutely? And give me a little bit of an understanding and the rest of the panel an understanding. What kind of specialty laboratory does that have to be sent to?

MAJORITY CHAIRMAN DIGIROLAMO: Rachel, come up.

DR. LEVINE: So all of the newborn screening in Pennsylvania is sent to PerkinElmer. They do all of our screening for Pennsylvania.

REPRESENTATIVE FRITZ: So the process would not be impeded, would not be held up? It's still a simple, straightforward process?

DR. LEVINE: For the initial testing, yes.

REPRESENTATIVE FRITZ: Okay.

DR. LEVINE: But there is subsequent testing. If there's a positive, there's subsequent testing that has to be done. The initial test is not particularly specific or sensitive.

REPRESENTATIVE FRITZ: And can you speak to the increase in cost of the test?

DR. LEVINE: So the issue of the initial test is not the cost. There is cost about secondary testing. But I want to emphasize the decision to have Krabbe currently be on the secondary list, so the follow-up list, so that as was mentioned, parents or physicians or health systems can do testing for all the lysosomal storage diseases if they're requested. I know that Penn State Hershey, as has been mentioned, does that.

So this was not a cost issue. This was an issue, again, following the recommendation of the national RUSP, the national committee, as well as our own committee had to do with the science of the sensitivity and specificity of the testing, particularly, how many false positives we would have, how many families would be notified that their child might have this obviously devastating and fatal disease and then have to have another blood test done and then have for a significant number of families have to go to a genetic center for further testing to diagnose one or two patients in which the treatment has not been published to be very successful.

So this was never a cost issue. This was an issue in terms of the science, and we have been following the recommendations of the national committee where Krabbe is not on the RUSP, as well as the TAB where it was recommended to put it on the secondary list but not to make it mandatory. It was never a cost issue.

REPRESENTATIVE FRITZ: Okay. Thank you.

MAJORITY CHAIRMAN DIGIROLAMO: Representative Miller.

REPRESENTATIVE MILLER: Thank you, Doctor. If you could, maybe that would be helpful.

Doctor, thank you for your testimony, as always. I apologize; I did have to step in and out. I just wanted to be sure. The New York report that you referenced --

DR. LEVINE: Sure.

REPRESENTATIVE MILLER: Did I get it straight that you're saying that they tested 1.9 million children, and at the end of it there were five positives, true positives that were found?

DR. LEVINE: That's correct.

REPRESENTATIVE MILLER: Okay.

DR. LEVINE: So they tested --

REPRESENTATIVE MILLER: Can I ask you this --

DR. LEVINE: One point nine million, 620 infants -- which I'm reading from the article -- 620 --

REPRESENTATIVE MILLER: Okay.

DR. LEVINE: -- infants were subjected to molecular analysis or DNA testing. Three hundred and forty-eight were referred for diagnostic testing to a genetic center. Five had Krabbe. One did not have treatment. Four had transplantation. Two died from transplant-related complications, and two survived with moderate to severe handicaps.

REPRESENTATIVE MILLER: Thank you, Doctor. Does the report say how many, of the children who were tested, came back as negative but were found to have --

DR. LEVINE: None.

REPRESENTATIVE MILLER: None. Okay. So the test works.

DR. LEVINE: If you go through all of the different -- the primary, secondary, and tertiary testing, the test will work.

REPRESENTATIVE MILLER: Okay. One point nine million, five positive, none of the tests that came back negative were proven to be incorrect?

DR. LEVINE: That's correct. That's correct.

REPRESENTATIVE MILLER: Okay. The test -- I mean, that's a pretty good margin. Wouldn't you agree? I mean, how many more -- let me ask you this. How big of a margin here do you need before you would say that the test works?

DR. LEVINE: I guess the test has a high specificity but not a high sensitivity. So that's medical terms.

REPRESENTATIVE MILLER: Okay.

DR. LEVINE: So when you say "works" meaning it's very unlikely to have a false negative.

REPRESENTATIVE MILLER: Right. But did it --

DR. LEVINE: However, you can have --

REPRESENTATIVE MILLER: -- didn't miss anyone?

DR. LEVINE: -- significant false positives.

REPRESENTATIVE MILLER: It didn't miss anyone?

DR. LEVINE: That is correct.

REPRESENTATIVE MILLER: Okay. And you went over earlier that the cost is minimal.

DR. LEVINE: The cost is minimal for the initial testing. As further testing goes by, there is significant cost, but this was -- as I would testify before, never a cost issue. That was not why we made our decisions.

REPRESENTATIVE MILLER: Are there any genetic predispositions that would, you know, sort of alert parents to a greater propensity of likelihood their child may develop this?

DR. LEVINE: If they had a previous child with Krabbe or if Krabbe had been in their family. Otherwise, no.

REPRESENTATIVE MILLER: Okay. So it's just direct history. There's no other connections --

DR. LEVINE: No.

REPRESENTATIVE MILLER: -- from the -- okay. I'm not intimately familiar with Representative Cruz's bill. We've heard the word mandatory. What is brought -- when children -- I remember my child when he was born, that was -- of course, you know, we went through this 10-plus years ago now. Do people talk -- are options given for testing with the pediatrician -- or, I'm sorry, optometrist? What is it? I don't even remember.

DR. LEVINE: The obstetrician.

REPRESENTATIVE MILLER: Yes.

DR. LEVINE: It could be the patrician, but newborn screening is usually done right when the baby's discharged from the hospital so usually a couple days later.

REPRESENTATIVE MILLER: Okay.

DR. LEVINE: If the child has to stay for a reason, it'll be done during the hospital stay.

REPRESENTATIVE MILLER: Is this routinely discussed --

DR. LEVINE: Yes.

REPRESENTATIVE MILLER: -- as an option?

DR. LEVINE: It's routinely done, and we -- again, Pennsylvania has an unusual system because we have the mandated tests and then we have a secondary list, as has been talked about. I've never seen that before, but that's the way Pennsylvania is. And different health systems will have different policies, but in terms of the possibility of further testing, we would recommend that doctors talk with their patients about what testing would be available. A lot of it depends what hospital or health system you're at and how their --

REPRESENTATIVE MILLER: Okay.

DR. LEVINE: -- testing works in their newborn center. But if a testing isn't recommended or done at that hospital, we would recommend that doctors tell the --

REPRESENTATIVE MILLER: Okay. So some doctors will talk about it with the parents and some will not?

DR. LEVINE: Well, we would recommend that all doctors talk about it with their patients.

REPRESENTATIVE MILLER: Right. That's your recommendation. I get it. I'm just trying to understand what's going on now. So I got your recommendation. Right now, are we just not sure if some doctors are talking, some are not? We don't know --

DR. LEVINE: It's hard for me to tell you what every doctor does with their patients.

REPRESENTATIVE MILLER: I'm not saying every doctor. I'm asking for a generalization as to what is brought up at these -- yes.

DR. LEVINE: So if you don't mind, the doctor from the Newborn Screening might be able to answer some of these questions.

DR. YUDELL: Thank you for your question. I think it's a really important question.

And as it exists now, there is very little education about the Newborn Screening Program that parents receive.

REPRESENTATIVE MILLER: Okay.

DR. YUDELL: They receive a pamphlet upon discharge from the hospital. There's a discussion with a nurse when the baby's wheeled away. Parents can opt out for religious reasons currently. But that's really it. There are concerns that I have as an ethicist about parents and moms in particular who are, you know, 24, 48 hours postpartum and having to absorb information like this. That is a concern, which is why I think getting back to the bill and the newborn screening fee, that money could be used to educate parents about all of the diseases on the mandated list right now, diseases on the secondary panel to allow parents to make informed decisions about what their children have to be tested for based on public health law and what their children could potentially be tested for. And I think that's --

REPRESENTATIVE MILLER: Okay. And I'll finish up because I know the Chairman's got to keep this moving.

You know, I would tell you that we had a bill that came up -- I think it was in my first session -- that had to do with information provided to parents upon a Down syndrome diagnosis. And it had its degree of controversy. You know, I know that some felt that the Legislature was intruding too much into a doctor-patient relationship by prescribing specific information that should be discussed.

I get it. Not all of these issues are as easy to deal with.

I guess my overall point, though, is that I've always leaned towards more information, not less, and I've always been concerned. As a parent who's had a diagnosis for their child, not as severe, but has had a diagnosis for their child, I remember what I didn't get. And I remember that feeling, and I remember sitting in that parking lot and crying with my wife as we tried to read the scribble that was given to us from the doctor as to what might be good for our child to do, and our needing to search Facebook, the Internet, call somebody's cousin to find information about what could possibly benefit our child.

Because as much as I appreciated a lot of the medical profession's input -- in particular, the nurses were always very helpful with it -- there were voids, massive voids that came about. Luckily for us, the diagnosis was not of a life-threatening situation but a life-lasting situation.

And so, you know, I appreciate the testimony today, especially your expertise, as well as the impact for both mothers being so brave to talk about it. I appreciate, first, Chairman Cruz for bringing it up and for Chairman DiGirolamo for letting us talk about it today. I look forward to hopefully considering some more options on this.

More information has got to be better. And like one of the moms that said with it, I'd rather have a -- well, my feeling of her testimony, I'd rather have a chance than no chance. Thank you.

DEMOCRATIC CHAIRMAN CRUZ: Dr. Levine, I have a question. When did the report come out?

DR. LEVINE: Which report?

DEMOCRATIC CHAIRMAN CRUZ: The New York version.

DR. LEVINE: This report is March 2016. And there is nothing in the medical literature about really testing or screening. Since that time, there was an article "Newborn Screening for Lysosomal Storage Diseases: Views of Genetic Health Care Providers." That was April 2016 in the *Journal of Genetic Counseling.* And they also recommended not to screen for Krabbe's because the lack of data about successful treatment.

DEMOCRATIC CHAIRMAN CRUZ: But New York City does testing for Krabbe's --

DR. LEVINE: New York State still does continue to test.

DEMOCRATIC CHAIRMAN CRUZ: The next speaker will be Aaron Kaufer.

REPRESENTATIVE KAUFER: Thank you, Chairman Cruz.

Thank you for your testimony, especially to the mothers that were here today. Thank you, really. I have to say I'm moved. I really am.

I have a couple of questions --

DR. LEVINE: Sure.

REPRESENTATIVE KAUFER: -- and I think the two people up at the microphones I think could answer them. Do we have a percentage of what the false positives are? Is there any percentage or --

DR. LEVINE: Well, so what we estimated is that in Pennsylvania we would probably have about approximately 8 to 900 false positives a year. So that was an estimate in terms of how many babies we have born and in terms of screening, but the estimate was that we would have approximately -- this is from Dr. Vockley -- about 8 to 900 positives, and we might have one positive child in a year. Now, that's just looking at statistics, but the estimate would -- I mean, whatever one out of -- the false rate would be significant, yes.

REPRESENTATIVE KAUFER: So you mentioned about also the secondary and tertiary testing.

DR. LEVINE: That's correct.

REPRESENTATIVE KAUFER: So out of that 8 to 900 hundred, how many would be weeded out with a secondary test?

DR. LEVINE: If you did a DNA test, probably about 6 to 700 would be weeded out --

REPRESENTATIVE KAUFER: And then you would be down --

DR. LEVINE: -- and then several hundred -- yes, they --

REPRESENTATIVE KAUFER: So then you'd be down to 2 to 300?

DR. LEVINE: That's correct. And then we estimated that several hundred would have to go to a genetic center for further testing at one of the six I believe, you know, genetic centers in Pennsylvania.

REPRESENTATIVE KAUFER: And so that would then bring it down to what you're saying would be statistically about one --

DR. LEVINE: About one a year --

REPRESENTATIVE KAUFER: Okay.

DR. LEVINE: -- we estimated might be positive for Krabbe's.

REPRESENTATIVE KAUFER: So I think the other thing you were referencing was that March 2016 study.

DR. LEVINE: That's correct.

REPRESENTATIVE KAUFER: And I think you said that out of the five people that were in that study two were successful in the treatment, right? Is that correct?

DR. LEVINE: So I'll read it from the study. One parent -- four underwent the bone marrow transplantation in the newborn period. Two survived with moderate to severe handicaps and two died from transplant-related complications over an eight-year period.

REPRESENTATIVE KAUFER: So that's a 50 percent success rate to some degree then?

DR. LEVINE: Of the four patients, yes, two -- although the two that survived still had moderate to severe handicaps.

REPRESENTATIVE KAUFER: Okay. Because I look at it this way, and maybe it's a slightly different perspective, but, you know, there's a lot of treatments we do all around the Commonwealth, and the success rate is not 50 percent. I would imagine there's a lot of things we do that are lower than that. And would the fact that the Affordable Care Act is the law of the land currently and that preexisting conditions are covered, that this would be something that would be covered under health care right now.

DR. LEVINE: Yes. Yes. It would covered.

REPRESENTATIVE KAUFER: So I just have to say I think the point was made that we want to educate people more so about these tests, but if we educate them about it, I think most people would say, yes, I want to have those tests done for my child. So it's almost, you know, we want to spend the money to educate them, and I'm thinking as any responsible parent, my child has a, you know, 1 percent or less than 1 percent. I want to know if my child has that because, you know what, there is a 50 percent success rate of treatment if it's identified.

And to me there's like two arguments going on here, and I think it's very important to distinguish what these two arguments are. One is we have this high number of false positives with the current testing that we have and there has to be more intensive training. And with time, technology will catch up, and I do --

DR. LEVINE: Yes.

REPRESENTATIVE KAUFER: -- agree with that. So that's the one argument is we have to filter through all these numbers. But the second argument is, well, the cost and treatment could be very high and might not be successful. But, you know, we have, you know, a Federal health care law for a reason, that this is covered under preexisting conditions. And I can tell you just from listening to the parents today, if given that opportunity, given that chance, I guarantee they would have taken that opportunity.

So I just want to say thank you. That's sort of where my mindset is at. And I think the parents today, you really left a compelling argument. And thank you for the courage and bravery to come up and talk about your personal story. Thank you.

DEMOCRATIC CHAIRMAN CRUZ: Anybody else?

Representative Warren.

REPRESENTATIVE WARREN: I believe the Representative's comments are very consistent with our passage just last week of the Right to Try legislation in that if this newborn screening presents a chance, an opportunity, it's one we should avail ourselves of.

Dr. Levine, you said a couple times that it's not a cost issue, and I'm not entirely clear. Can you succinctly explain exactly what the issue is?

DR. LEVINE: The issue in terms of why it was placed on the secondary list and not the primary list was the science was that it is not recommended by the national committee that forms the RUSP, and it was not recommended by our own committee, our own Newborn Screening committee to add to mandatory to screening because of the concerns about the poor sensitivity of the test meaning how many false positives there would be, how many families that would have to go through different layers of testing to have one -- and maybe to make one diagnosis.

And it's not the cost of that. It is never the cost. And to your comment, it was never the cost of treatment. It was how many families would be put through that suffering of maybe thinking that their child had Krabbe's, in fact going to a center and then getting an appointment with a center and then, you know, having to wait for the test results, et cetera.

But then also the lack of scientific evidence about the success of the treatment, meaning -- so those are the two requirements that the national board looks at. They look at can the testing be done successfully and is the treatment successful. And the published results of the treatment are not very good. We have heard some anecdotal evidence about some better results but nothing that's been published in the literature, and we look at evidence-based medicine about whether the treatment would be successful.

So we are following the recommendations of both the national and our Pennsylvania Newborn Screening committee, which does not recommend at this time adding Krabbe. Things change. I mean, Pompe and MPS I were added over the last two years so those are part of the mandated screening now. It's on the RUSP. Our committee has approved it and it is there. Krabbe has not been approved by the national or the Pennsylvania committee. It has not been recommended to add because of the science. It was never any issue about the cost of screening or the cost of testing or the cost of treatment.

MAJORITY CHAIRMAN DIGIROLAMO: Representative

Madden.

REPRESENTATIVE MADDEN: Hi. Thank you, Mr. Chairman.

And I would like to thank Vicki and Lesa for your compelling and really devastating testimony.

I am the grandmother of a six-month-old grandson, and I can tell you I would want to know everything, you know, given the opportunity. And something that Lesa said that brings a question to my mind is that they were fortunate enough to be at a hospital that did test for Krabbe. Do you believe that we as a State would be doing a better service to our newborns and their parents if we had a uniform testing panel that every hospital tested for the same diseases at birth so that by the misfortune of being at one hospital you aren't being tested if you knew?

And I would just like to add that, you know, I'm a freshman; I haven't been here that long, but the more I'm here, the more disheartened I am by the fact that Pennsylvania is at the bottom in so many areas. And I'm wondering at what point do we lift ourselves up and say why are we relying on a New York study? Why aren't we doing those studies? Why aren't we treating those babies and using those results to be, you know, the premier State or premier, you know, leader in finding out the cures.

And, you know, I'm sure that if a mother knew or parents knew that their baby was going to die regardless, they would be willing to be part of a study that would give them hope and would give hope to future parents.

So I guess that one question in there was do you think we would benefit from a --

DR. LEVINE: Yes.

REPRESENTATIVE MADDEN: -- uniform panel of tests?

DR. LEVINE: I think that we would benefit from a uniform panel as opposed to the mandated one and then the supplemental one. And I think that it needs to be part of the newborn fee as opposed to something that the State pays for. So you're asking me a separate question than the issue with Krabbe. So yes, we believe it would be better to have one panel.

REPRESENTATIVE MADDEN: So what do we have to do to make that happen?

DR. LEVINE: Legislation.

REPRESENTATIVE MADDEN: Okay.

DR. YUDELL: Can I --

DEMOCRATIC CHAIRMAN CRUZ: By the way, my legislation will do that.

DR. YUDELL: Can I supplement that answer if that's okay?

REPRESENTATIVE MADDEN: Sure.

DR. YUDELL: So what I think we'd like to see happen as a committee is to someday add Krabbe. And I think that, you know, everything that's been said here is very compelling. I think our concern remains that there's one study looking at this that shows both good and bad outcomes. And it would be great if we had more data.

My concern, though, in framing it as a research issue in the State of Pennsylvania is that that could trigger the public to be concerned about whether research was being conducted on their babies. So I think that, you know, if Krabbe is to be added, it should not be framed as a research question because I think that would --

REPRESENTATIVE MADDEN: So if I framed it as a --

DR. YUDELL: -- raise issues of --

REPRESENTATIVE MADDEN: -- treatment question, would that be better?

DR. YUDELL: Well, I mean, I think it's a public health concern and that we want to provide support to parents and babies in Pennsylvania to, you know, get the best interventions and treatments as early as possible to provide the best outcomes so --

REPRESENTATIVE MADDEN: Okay. Thank you.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Representative Nelson.

REPRESENTATIVE NELSON: And thank you. Sorry for the additional questions I'm trying to do my best not to ask but I just have to climb into the ring.

Going back in time, help me to understand why the original bill that was passed by the Chairman, the House, the Senate, signed by the Governor wasn't actually implemented? Because it's a frustration that if we have constituents -- and it's a difficult process to be able to bring an idea to law but then to feel subverted on the backside that, well, if for business issues or for the financial side -- I mean, as a parent of six, I would much rather know if there is, you know, a condition that they may or may not survive as an issue of choice.

But what drove the absence of implementation of the original bill?

SECRETARY MURPHY: I'll take responsibility for that. So as Secretary of Health, if we go through the process, the Secretary of Health can implement the test according -- the law says that the Technical Advisory Board can advise the Secretary of Health, and that is the way the law is written. And I would encourage you to continue that work with the Technical Advisory Board because they are the scientists; they are the experts.

Again, to echo what Dr. Levine said, it was never, never a cost issue. It had nothing to do with the budget. The decision was made by accepting the recommendation of the Technical Advisory Board. So the Technical Advisory Board in December -- we came in in -- I believe the dates if I'm correct it was October of 2014 when the legislation was passed. When we came in in January of 2015, we began to do due diligence because this was on our implementation list. So --

REPRESENTATIVE NELSON: I think maybe the concern is that, yes, the Technical Advisory Board plays a crucial role, but if we pass a law and the Governor signs it -- you know, some of these babies in 2016, '17 may now survive even at a better rate because we would have actually started to do something. I think it's the inaction that, you know, gives us pause because -- and it makes us less trusting of future legislation allowing the fact that even if we go to that 1 percent situation, that, hey, even though we work and we pass and we meet the needs that the people are calling, you know, we may not get it actually across the finish line because we could have babies living now that maybe aren't going to in 2018 and 2019.

SECRETARY MURPHY: And I think -- and I say this with all due respect. In terms of the implementation, we do have Krabbe testing available in Pennsylvania. I think that's something -- it is because of the difference in the mandatory and optional panel. But I do want to be clear that Krabbe testing is available in Pennsylvania.

I also want to be clear, back to Representative Miller's question about physicians educating parents, I think what you're seeing in terms -- if there are physicians that are not discussing this secondary panel option is because they believe in their medical decision-making that the benefits of doing that testing does not exist to the full extent because a physician in counseling a parent on newborn screening, these, again, are people who are dedicated to producing positive outcomes.

So I think what we have here, and not to diminish -- I am as moved, believe me, as you all are and this heart-wrenching testimony, and I wish that we could save everyone's child. I'm a mother myself. But I think what you're seeing, if you're not seeing the level of Krabbe testing by physicians is because they don't believe it's in the best interest as of yet of their patients.

Now, as Dr. Levine pointed out and Dr. Yudell pointed out, we met with Dr. Vockley 48 hours ago or maybe that was --

DR. YUDELL: It was last --

SECRETARY MURPHY: Oh, it was last week.

DR. YUDELL: About five days ago.

SECRETARY MURPHY: Sorry, my time is -- but very recently to say I take this very seriously. This was not a decision that we made lightly. This was something -- I think the Representatives that have worked with us understand our commitment to the health of Pennsylvanians. This was not something that I made this decision lightly. I did extensive due diligence talking to the national experts, as well as our technical advisory committee.

I fully support a uniform panel. I fully support mandatory testing according to the national panel and the recommendation of our Technical Advisory Board. So I assure you that this was not made in any intention of disrespect. It was made by research and actually listening to the Technical Advisory and the national panel.

Again, we support -- I agree, going back to Representative Madden's point, every Pennsylvanian deserves the same shot at, you know -- so it shouldn't matter where your baby's born as to what tests you have. Every Pennsylvanian parent and child deserves to have the same chance.

And I think that, you know, Representative Cruz has done extensive research with this. He has talked to the experts. I hope that the work of the technical advisory committee is I think really recognized and highlighted in this. I think to the prior comment about lifting Pennsylvania up, I think we do have to lift Pennsylvania up. And I think by engaging the experts and developing legislation that protects all Pennsylvanians is the way to do that.

But I want to be perfectly clear that this was not a decision that I took lightly.

REPRESENTATIVE NELSON: Thank you. Thank you, Mr. Chairman.

DEMOCRATIC CHAIRMAN CRUZ: Just to echo on what Representative Nelson says, this is the law. No one's above the law. And if the law is the law, then we have to apply it to everyone, and we're not doing that. Unless the parent says to the doctor I want my child tested for this, people are not educated. People don't know that you can get this done automatically.

So how do we move forward? We've gone through all the panels. We've gone through all the decisions. We've gone through everything that even with all that, we've gone and made it into law. It passed the House, passed the Senate, the Governor signed it, but we're not implementing. Who do we hold responsible for not implementing the law?

SECRETARY MURPHY: I think as my understanding and our legal review representative is that the Technical Advisory Board is -- the Secretary of Health is given the authority to implement newborn screening. You know we've had this discussion over the last year. I think the way to move forward is the way that you are proposing in terms of a uniform panel. Again, I think every Pennsylvanian deserves to have the same quality of care regardless of where they're at. I do not believe by the evidence that we talked about today that physicians are being negligent or hospitals are being negligent. I believe that they are offering what they believe to be the evidence-based medicine.

I do think that the advantage of following the RUSP, which is the national benchmark about what we should be doing, I think by following the RUSP, I think Pennsylvanians, we will move to the forefront of what is being advised, and we will be able to improve the offering to parents and children in Pennsylvania.

DEMOCRATIC CHAIRMAN CRUZ: Gene, when we're done here after all this testimony is done, I would love to have the two mothers come up and ask any questions from us --

SECRETARY MURPHY: Sure.

DEMOCRATIC CHAIRMAN CRUZ: -- and for us to ask from them because we're trading here information that is going to make Pennsylvania the best that we can. So I just wanted to put that, Gene, and I apologize.

SECRETARY MURPHY: And I would assure you that Dr. Levine and I and everyone at the Department of Health wants the same for everyone in Pennsylvania.

MAJORITY CHAIRMAN DIGIROLAMO: Representative?

REPRESENTATIVE NELSON: I just didn't -- in circling back, I mean, I think the true disconnect is -- you know, these mothers are caught in the balance where this is a really good example where the policymakers are ignoring the law and the Legislature -- you know, a law was passed, and the policymakers are not following -- and I understand that doctors may choose or feel, and I truly respect the authority of your position, but the law was passed. The House, the Senate, the Governor placed it in, and the people placed us here. And the people want that law implemented, and you're not doing it, Madam. And that is I think a theme that is a much larger concern point.

And I understand and respect the level of knowledge, outstanding testimony and fact-based 2016, but the bigger issue is as we move forward with legislation that's stemming from the people of Pennsylvania, it should be implemented. And if there is a disconnect, then that would circle back and we would adjust the legislation. And that just doesn't seem to happen in this situation.

SECRETARY MURPHY: And it did in terms of shortly after I arrived -- and I think the Department of Health has been engaged fully with Representative Cruz's office and staff. So this was not something -- this is not the first time we're discussing this issue. As soon as the issue came that there was a disconnect and that the Technical

Advisory Board did not support the -- it did not support Krabbe testing and we put it on the secondary list, the second that -- I think Representative Cruz would attest to that -- we have been actively engaged in this discussion. This is not something that we just decided now to do and didn't discuss it further.

So, you know, I certainly respect the legislation, and I respect everything that Representative Cruz is doing. And we will continue to work with he and his staff and the Technical Advisory Board on moving forward.

MAJORITY CHAIRMAN DIGIROLAMO: Representative Kaufer for follow-up.

REPRESENTATIVE KAUFER: All right. Just a couple of questions. And I honestly have no idea; I haven't heard any of this discussed yet. What is the likelihood of a child living with Krabbe to adulthood?

DR. LEVINE: There are a number of different forms of Krabbe. There's early-onset and late-onset. For early-onset, it's zero. For late-onset, then -- I'm not an expert, but they can live longer. But for the tragic cases that you've heard before and with other babies with early-onset Krabbe's, there's no chance.

REPRESENTATIVE KAUFER: Okay. Because --

DR. YUDELL: Without treatment.

DR. LEVINE: Excuse me?

FEMALE SPEAKER: Without treatment --

DR. LEVINE: Without treatment --

FEMALE SPEAKER: -- there's no chance.

DR. LEVINE: -- that is correct.

REPRESENTATIVE KAUFER: Okay. So with treatment, though --

DR. LEVINE: The literature is as you saw --

REPRESENTATIVE KAUFER: Right, 50 percent.

DR. LEVINE: -- two babies have severe -- we don't know what their life expectancy will be, but two babies are, according to the literature that I've shown you, have moderate to severe impacts, but that as of the publication, they are alive.

REPRESENTATIVE KAUFER: Okay. Because one of the arguments that was made today was that parents will be suffering not knowing if their child might have it or might not have it, you know, because it seems like if the treatment is not done, you will guarantee that the child will die. And that certainly seems to be quite some suffering in and of itself.

DR. LEVINE: Of course.

REPRESENTATIVE KAUFER: And so there's a couple arguments that were just made today that I just -- I mean, there's three or four different arguments that I just think are swiss cheese. I mean, they are really full of holes. And I appreciate where you're coming with some of this stuff, but, you know, if you break down each separate argument that has been made today, I think there are really some massive holes in each argument that was laid out today.

So thank you, Chairman, and thank you for bringing this to my attention. This was an issue I was not aware of, and I appreciate the fact that we're talking about this today.

MAJORITY CHAIRMAN DIGIROLAMO: Representative Miller for follow-up.

REPRESENTATIVE MILLER: Okay. I got to tell you, I appreciate -- first off, I appreciate this Committee because we get the chance to see some new people, so get to meet some new people. Hey, how are you doing? And I appreciate hearing Representative Nelson's comment to focus on an issue that I had glossed. And, you know, first off, I apologize for anything with this, but I guess I'm just a little surprised. So just so I got it straight, October 14 we passed a law, Governor signed it, you then have a -- tell me the board again.

SECRETARY MURPHY: Previous law states that the Technical Advisory Board --

REPRESENTATIVE MILLER: Okay. So Technical

Advisory Board --

SECRETARY MURPHY: -- on newborn screening.

REPRESENTATIVE MILLER: -- what do they do?

SECRETARY MURPHY: In fact, Dr. Yudell is the co-chair. He can explain.

REPRESENTATIVE MILLER: Co-Chair.

DR. LEVINE: Would you want to discuss?

REPRESENTATIVE MILLER: Okay. What do they do?

DR. YUDELL: So we meet quarterly to both review the current tests to see their success and their failures --

REPRESENTATIVE MILLER: Right.

DR. YUDELL: -- and we also discuss adding new screens and reviewing new screens to the panel.

REPRESENTATIVE MILLER: And you make -- I imagine when you're reviewing or thinking about new screens, you make recommendations to go to --

DR. YUDELL: The Secretary of Health.

REPRESENTATIVE MILLER: Secretary of Health.

DR. YUDELL: Based --

REPRESENTATIVE MILLER: Okay.

DR. YUDELL: Yes, based on specific criteria.

REPRESENTATIVE MILLER: Can I ask any of you this knowing that I'm unfamiliar with this technical board thing here? Somewhere does it say that the laws that we pass are optional for you to help implement?

SECRETARY MURPHY: In this case, the Technical Advisory -- the legislation that issued the Technical Advisory Board -- because, believe me, we have looked into this at the time and since --

REPRESENTATIVE MILLER: I do hear somebody say believe me a lot in D.C., so as soon as you say, believe me --

SECRETARY MURPHY: I won't say believe me or honestly --

REPRESENTATIVE MILLER: Go ahead.

SECRETARY MURPHY: -- yes, we won't use those two words, believe me or honestly.

REPRESENTATIVE MILLER: Yes.

SECRETARY MURPHY: I think that -- well, I know that the legal opinion that was provided to the Department of Health was that the Technical Advisory Board can recommend to the Secretary of Health and the Secretary of Health can make the decisions based on the Technical Advisory Board.

REPRESENTATIVE MILLER: Okay.

SECRETARY MURPHY: I do want to point out, too, we're talking about this in 2017. At the time, this wasn't just a simple discussion. Like the Technical Advisory Board, if Dr. Yudell will share, felt very strongly about this issue. It wasn't --

REPRESENTATIVE MILLER: Right. And I would take it that let's assume that he's told you right, let's assume that, you know, he's totally right with it, I guess what I'm just trying to get to is that you're saying that you have a legal opinion that says regardless of what the Legislature passes and regardless of what the Governor then signs, regardless of what becomes law, you have your own type of negate or veto stamp over a public health law because of your position?

SECRETARY MURPHY: In this specific situation.

DR. YUDELL: Because --

REPRESENTATIVE MILLER: I'm sorry, I just want to understand. This specific you mean related to Krabbe?

SECRETARY MURPHY: Related to newborn screening.

REPRESENTATIVE MILLER: So newborn screening you have the complete decision on whether or not to do something?

SECRETARY MURPHY: Yes.

REPRESENTATIVE MILLER: So we're wasting our time. It's irrelevant for us to do this stuff because you decide. I mean, isn't that it? I mean, what are we doing here?

SECRETARY MURPHY: No, I really don't think that that's the case, Representative. I think that what you're doing is extremely important. I can tell you that in --

REPRESENTATIVE MILLER: For what?

SECRETARY MURPHY: -- this situation the public policy opinion across the country by experts -- and we made Krabbe available. I think what --

REPRESENTATIVE MILLER: Ma'am, you're not -- thank you. And I apologize because I don't want to take all your time. First off, I'd love to see the opinion, okay? So whatever --

SECRETARY MURPHY: Sure. We will --

REPRESENTATIVE MILLER: -- the opinion, I would love to see.

SECRETARY MURPHY: -- be happy --

REPRESENTATIVE MILLER: I got to tell you, just as -- you know, I'm shocked. I'm shocked. I've never known, hey, maybe it's something, hey, I just need to know. Each Secretary has a couple things that they can say, bam, forget it. And maybe that's the way it is. It could be that I'm uninformed and I just didn't catch that. So I appreciate you mentioning it. But I just have never known -- like I don't care what Utah is doing. You know, in my opinion, if Pennsylvania Legislature comes up and this is what comes up with it and the 49 other States say they don't like it, then I don't represent Arizona. I don’t represent New York, you know? And I get it. We want your opinions -- your opinions, we rely on your informed opinions, but at the end of the day, as far as I knew, there's only one body that makes law and there's only person who could veto that or not, and I thought that was the Governor.

DR. LEVINE: So the issue that came up, it has to do with a legal interpretation of the original law versus the law that was passed in 2014. And I forget the date but --

REPRESENTATIVE MILLER: Okay.

SECRETARY MURPHY: 2008.

DEMOCRATIC CHAIRMAN CRUZ: 2008.

DR. LEVINE: Okay. But there was a 2008 law that in terms of the Newborn Advisory Committee and newborn screening and the role of the Secretary -- so this was the legal opinion that we were given in terms of the interpretation of how the 2008 statute would be implemented. So it's not that we are ignoring laws. It's that it's a legal interpretation about the previous law from 2008.

REPRESENTATIVE MILLER: Listen, as I said, I will look forward to being educated about that. Thank you. I never heard about some conflict with something to do with 2008 law, so I appreciate it. I guess my closing thought on this at all is that I don't believe that any Secretary from any Administration should be overriding any part of legislation.

Now, I'll look to be educated on the topic and then we could have another discussion if need be, and maybe I'm just wrong. But as I said, it is something that shocks me from my understanding otherwise of how things are supposed to go.

Thank you, Mr. Chairman, for letting me learn.

DEMOCRATIC CHAIRMAN CRUZ: Thank you. And for my Members on both sides, Democrats and Republicans, I created a piece of legislation to remove the power away from your panel because no one's above the law, and the law's the law. We make the final decision and you have to implement it. So I'll be circulating that again. This is why this public hearing was held so that everybody would be informed. And I thank you. You know, I don't want to bash any one of you. You're doing your job. Thank you.

But we legislators are the ones that legislate law, and I will do whatever it takes to give newborn children that chance to live. So thank you.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Seeing no further questions, Representative Cruz had asked if the two moms could come up, Vicki and Lesa, maybe see if there's any questions from the Members.

MS. BRACKBILL: Before that, I have a few things

I'd like to clarify, things that I've heard throughout the hearing that are not entirely accurate. The first thing is about the hospitals not screening for it. I've spoken with several hospitals throughout this process because even though I may be "just a mother" and not a doctor, I have lived and breathed Krabbe for over two years now and this is my passion because no one should have to go through what we've had to go through.

Right now, the current policy is that any mother in Pennsylvania who asks a hospital to test her baby for Krabbe, it's supposed to be done. But I can tell you from moms who have followed my daughter's story, it's not being done. Hospitals also don't understand this situation. They say, oh, it's law so it's being done. So there's a massive education effort that needs to happen because this has been reported back firsthand to both Vicki and myself from our followers.

The other thing is that they keep saying it's not a cost issue, but I've been to the last four Technical Advisory Board committee meetings, and every single test that's discussed about making mandatory, that's the first thing they say. It will cost this much because the State currently has to pay for it. And I've heard them even reference that to Krabbe. So I really do believe that money has played a portion in that.

Also, four States currently screen, not three, and two more are coming onboard this year, including Louisiana.

Also, just because it's not published doesn't mean that it doesn't exist. I can name so many transplant success stories that are going on not only at Duke but at Children's Hospital of Pittsburgh, which is where both of our daughters were seen. So many transplant success stories, I can show you pictures, names, their parents would talk to you. Dr. Kurtzberg from Duke would talk to you. The treatment works. If it is caught at birth -- like they just in the last two months, they caught somebody in Missouri and in Kentucky and they were both transplanted. They are both home, zero signs of Krabbe. Owen from Tennessee, zero signs of Krabbe. The treatment works.

And about the testing, I don't know if you guys know this, but 1 in 125 people are carries of Krabbe. I'm from California. I moved to Pennsylvania and I married a Pennsylvania man, and we're both carries of the same genetic mutation. This can happen to anybody.

Between 2013 and 2015 I know of at least five babies in central Pennsylvania who are diagnosed with Krabbe and have all died. This is very prevalent in Pennsylvania for some reason, and we have to do something to stop it.

Also, even just knowing that you're a carrier, which I believe is what the study is referring to as false positives. I can't remember the number because it's been a while since I looked at that study, but even just knowing that you are a carrier can help eradicate this disease. If I had known, I would have had my husband tested before we got married or when we got married so that we knew, oh, hey, you're also a carrier; we shouldn't do this. But we didn't know, and knowledge is power.

Also, if we aren't screening and we aren't treating, how is the data ever going to improve? How are we ever going to be able to perfect this screening or the treatment when there's only four States currently testing. There are seven others that have legislation in place that have not implemented because of cost.

Also, in regards to the New York study, one of the two survivors that was mentioned that has severe to moderate to disability, I've met him. He came to the Hunter's Hope symposium last year, along with many other transplant survivors. He is thriving. He goes to school. He is the cutest little kid, and he is living life. So even though he may have disability, that doesn't mean that he is not alive. And his mother even gave her story at the symposium about how thankful she was that she had the opportunity for treatment because New York State screened. He was the very first one that was transplanted because the New York State program.

And the way that we as parents feel about this is that we would so much rather be told that our child was just a carrier or that it may have been a false positive than to find out that it's too late because here's the thing about treatment -- and we can say this because we've been here -- you might die because of a transplant because it is a big deal, but they're going to die without it. And so as parents, we want to try. And I believe that, yes, parents could have the opportunity to opt out as some of those in New York did, but I can tell you that my husband and I would not have.

If you have any other questions, then you can ask them, but I believe Vicki wants to say some things as well.

MS. PIZZULO: You said everything. You did it all.

MS. BRACKBILL: I took notes.

MS. PIZZULO: I was just getting -- from a mother's standpoint, I heard the doctors stating that they would get false positives and that would just make the parents so stressed about getting the false positive. Coming from a parent who had a Krabbe child, I would have traveled around the world to find out if it was a false positive.

I visit my daughter at a grave on holidays, during the week. She's at a grave because Pennsylvania does not test. False positive, that's just ridiculous. Coming from a parent, I'd rather have a false positive and know.

MS. BRACKBILL: Yes, absolutely.

MS. PIZZULO: If it's not about money, then what's it about? These are out children.

And that was it. And then you talked about the hospitals. Not all hospitals are -- they keep on mentioning secondary testing. They're not all doing it. Nobody knows about it. When you're --

MS. BRACKBILL: Or they assume that it's already being done.

MS. PIZZULO: Yes. And if you're in a hospital and you're giving birth, you're not thinking about what your baby is being tested for. You're thinking you have a healthy child and the hospital is going to do what it's supposed to do. They're going to give you the tests that you need to have. So let's not like pat ourselves on the back because we're doing a secondary testing because that's not working.

MAJORITY CHAIRMAN DIGIROLAMO: Representative Charlton.

REPRESENTATIVE CHARLTON: Thank you, Chairman.

Vicki and Lesa, thank you both for your courage of being here today and speaking to us.

I have a problem with the comments that we heard before that, you know, their success rate was tempered by the fact that the two who survived the surgery had moderate to severe disabilities. I have a daughter with a disability, wouldn't trade her for the world.

MS. PIZZULO: Right.

REPRESENTATIVE CHARLTON: So I have a hard time believing that this surgery is not worthwhile because there's not 100 percent success rate. You know, I'm going to ask the question and I think I know the answer already and I think everybody in this room knows it but I want it on record. If you had the option for the surgery knowing that your child may have a mild disability or a severe disability afterwards but you'd get to take that child home, would you do it?

MS. BRACKBILL: Absolutely 100 percent.

MS. PIZZULO: Yes.

REPRESENTATIVE CHARLTON: Thank you.

MS. BRACKBILL: And according to Duke, the success rate's actually been more like 90 percent. This may not be published, but it's fact.

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Okay.

Seeing no further questions, I want to thank the both of you. Do you know each other?

MS. BRACKBILL: Yes.

MS. PIZZULO: Yes.

MAJORITY CHAIRMAN DIGIROLAMO: You've met before, right?

MS. BRACKBILL: It's a tight community.

MS. PIZZULO: Yes.

MAJORITY CHAIRMAN DIGIROLAMO: Well --

MS. PIZZULO: It's really rare but there's a lot of us. There are a lot of us.

MAJORITY CHAIRMAN DIGIROLAMO: I know it's been said before about how courageous you are, but thank you both for being here really. We really appreciate your compelling testimony.

And, Vicki, I know I've known you for a long, long time, you and your mom --

MS. PIZZULO: Yes.

MAJORITY CHAIRMAN DIGIROLAMO: -- and, boy, little Hannah caused quite a stir there --

MS. PIZZULO: Yes, she did.

MAJORITY CHAIRMAN DIGIROLAMO: -- in our area there for the longest time. Yes. A lot of people were in love with her. Yes.

MS. PIZZULO: Yes, right away like, just, I don't know. It just happened so fast. It was just --

MAJORITY CHAIRMAN DIGIROLAMO: Okay. Yes.

MS. PIZZULO: And just one more thing. I just want to add to the stress what we dealt with every single day. We literally did not know if our children were going to die that day. They struggled breathing. Krabbe takes their ability to live away. They just lay there. They don't smile. They don't laugh. They are blind. They lose their hearing. They can't move. They have muscle and nerve pain damage. You'd wake up and pray to God that your baby would be alive by the end of the day. I would take a false positive stress over that stress every single day keeping my baby and Victoria alive. They would be here with us right now.

MAJORITY CHAIRMAN DIGIROLAMO: We --

MS. PIZZULO: And I just --

MAJORITY CHAIRMAN DIGIROLAMO: We got the point. And I think you've got a lot of support to do this on the Committee, I mean, everybody I heard.

And again, I want to thank my Democratic Chairman, Representative Cruz, for getting the bill passed. And I remember a football player from the Buffalo Bills --

MS. BRACKBILL: Jim Kelly.

MS. PIZZULO: Yes.

MAJORITY CHAIRMAN DIGIROLAMO: -- Jim Kelly, I think he's lost a child to the same thing --

MS. BRACKBILL: Yes.

MAJORITY CHAIRMAN DIGIROLAMO: -- and he went around the country, is actually here in Harrisburg. Representative Cruz had him here I think two times --

MS. BRACKBILL: He's a wonderful support to all of us.

MAJORITY CHAIRMAN DIGIROLAMO: -- calling for the testing here in Pennsylvania. So let's hope we do it. Okay? Anybody else?

I would like to just make an announcement for the Members. I'm not going to adjourn this hearing today. I'm going to just recess. There's a possibility we might come back and vote that bill maybe at the break, the first break, so I'm just going to recess the meeting and the hearing right now. And thank you again, everybody, for being here.

(The hearing concluded at 1:14 p.m.)

I hereby certify that the foregoing proceedings are a true and accurate transcription produced from audio on the said proceedings and that this is a correct transcript of the same.

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