TODD NOVAK

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P.O. Box 8953
Madison, WI 53708-8953

DATE: March 17, 2021

RE: Testimony on Senate Bill 194

TO: The Senate Committee on Health

FROM: State Representative Todd Novak

Thank you, Chairman Testin, and members of the Committee on Health for holding this public hearing on Senate Bill 194 (SB194), which adds Krabbe disease testing as a required test during newborn screening.

Newborn children are screened for congenital and metabolic disorders. According to the state's Department of Health Services (DHS) there are 46 different disorders that are currently tested. Existing law requires healthcare workers to take the steps involved for testing newborns before an infant is discharged or within one week if the infant was born in a non-hospital setting. The Wisconsin State Laboratory of Hygiene conducts the lab work and results are then shared with the family and their physician. Newborn testing is paid for by a fee imposed by DHS. Current law also allows for those with religious or personal convictions an exception from newborn screening.

A family living in my legislative district had a child diagnosed with Krabbe disease. In their case screening was not conducted at birth and they only found out months later. When it comes to detection time is of the essence. In response, I introduced legislation in February of 2015 that would have provided for the testing of Krabbe disease at birth. That bill did not advance to become law. The issue remains as newborns are not being screened for this disease at birth.

SB194 would require globoid cell leukodystrophy, also known as Krabbe disease, to be tested for as a part of the newborn screening process. Currently, DHS has the ability to promulgate rules for the testing of additional congenital and metabolic disorders. In the case of Krabbe disease this has not been done. While Krabbe disease has no cure, early detection of the disease at birth enables parents to seek treatment for their children. As a result they are then able to live longer and fuller lives. This bill also requires DHS to review on a biennial basis the appropriateness of additional lysosomal storage disorders for screening. After this review, the Department will then be able to promulgate rules for the inclusion of additional newborn screening if appropriate.

SB194 makes a simple change that I believe will positively impact children affected by this disorder. I want to thank you for your consideration of this proposal.

Testimony from Kevin and Judy Cushman

Dad's Testimony:

My name is Kevin Cushman and I am the father of Collin Cushman who passed from Krabbe.

I'd like to start by asking, How many of you have children?

How many of you remember where you were when you found out that you were going to be a parent for the first time?

Did you just get goose bumps?

What a happy and exciting time!

Now I'd like you to picture your child at 13 months of age....

Imagine yourself sitting on the floor holding your child as the phone rings....It's the Genetic Doctor calling....How do you think you'd feel hearing the doctor tell you that your son has a terminal disease. And that the life expectancy is 13 months to 2 years.

Collin was 13 months old when he was diagnosed. Did that mean that I only had a few months before losing him?

Krabbe affects each kiddo differently. Some parents have to make end of life decisions before their child reaches 2 years old. Decisions such as stopping feeds because their child was aspirating or taking them off oxygen because it was causing too much discomfort. And the result is watching their child slowly wither away until they pass.

But Collin beat the odds. He was a fighter and stronger than I could ever hope to be. He was 8 years and 18 days old when he passes. But it didn't come without difficult times.

It costs at least $700,000 a year to care for one affected child. Collin was diagnoses on January 6, 2012 and passed on January 6, 2019. That's 7 years. Thankfully Collin's medical expenses were covered by Medicaid.

Collin endured so much in is short life. Severe tone issues, vest treatments, nebulizer treatments, feeding tube replacements, an ambulance ride, at least a dozen hospital stays, irritability, being on oxygen or having to be suctioned frequently because he couldn't manage his own secretions. And this just a very small list of what he dealt with on a daily basis.

As a parent, imagine waking up every single day as I did, wondering if this was going to be the day that Krabbe was going to take your son's life.

Imagine caring for your child 24/7 for 8 years and 18 days and knowing more about his disease than you'll ever know about your son.

It's a horrible feeling knowing that as a parent there is nothing you can do for your child but to keep him as comfortable as possible as you wait for the disease to take his life.

But Collin took all that life gave and all that Krabbe took from him in stride. But he shouldn't have had to deal with all this. No child should.

Now I'd like you to imagine how you'd feel finding out that had Wisconsin been screening for Krabbe, that there was a treatment! A treatment that could have given not only a longer life but a better quality of life. A treatment that could give you chance to have a conversation with your son. A chance to know your son. There is a 10 year old boy right now (which is how old Collin would have been) living in Oregon that received the transplant is disease free.

Wisconsin needs to be screening for Krabbe. Because there will be another Krabbe child born in Wisconsin and this treatment would give those parents two things that Judy and I never got. A choice and hope.

Shortly you'll hear from the Thom's family and they'll share with you exactly what this means. You'll also hear more about the treatment from Dr. Kurtzberg and about the screening cost from Dr. Gelb

It was too late for Collin. And we don't want another kiddo to have to go through what Collin went through or parents to have to go through what we went through and will be going through for the rest of our lives. That's why we are here.

You have a great opportunity in front of you today. A chance to be proactive because there will be more kiddos born in Wisconsin with Krabbe. We are asking that you vote in favor of SB-194.

Thank you.
Kevin Cushman

Mom's Testimony:

Collin's life impacted my life, now it's time for him to impact yours, and impact babies being born in Wisconsin. He brought us here today to bring Krabbe to the forefront.

Let me give you an idea of what part of his day was like. Collin's schedule was very routine; varying it could cause many health issues that could put him in the hospital. He required 24/7 care. He couldn't take care of himself at all. He received medicines six times a day. He had five feeds.

Having a feeding pump continually on him caused spit ups. We found that pushing in his feeds by 15 ml every five minutes enabled him to tolerate it better. That means it took close to an hour for one feed. He was held a lot because of burps and suctioning. He was suctioned, on average, five times per hour. He had three vest and cough assist treatments and two nebulizer treatments. This allowed his secretions to be suctioned rather than staying in his lungs.

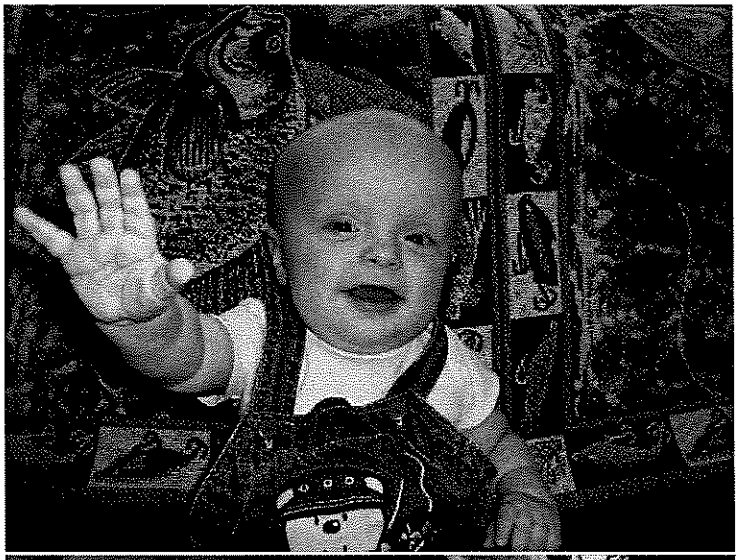
We did range of motion exercises with him. He didn't do any of this on his own. Afos and knee immobilizers had to be put into his stander. It was never easy to add anything to his day, because it all needed to be done every day and consistency was important.

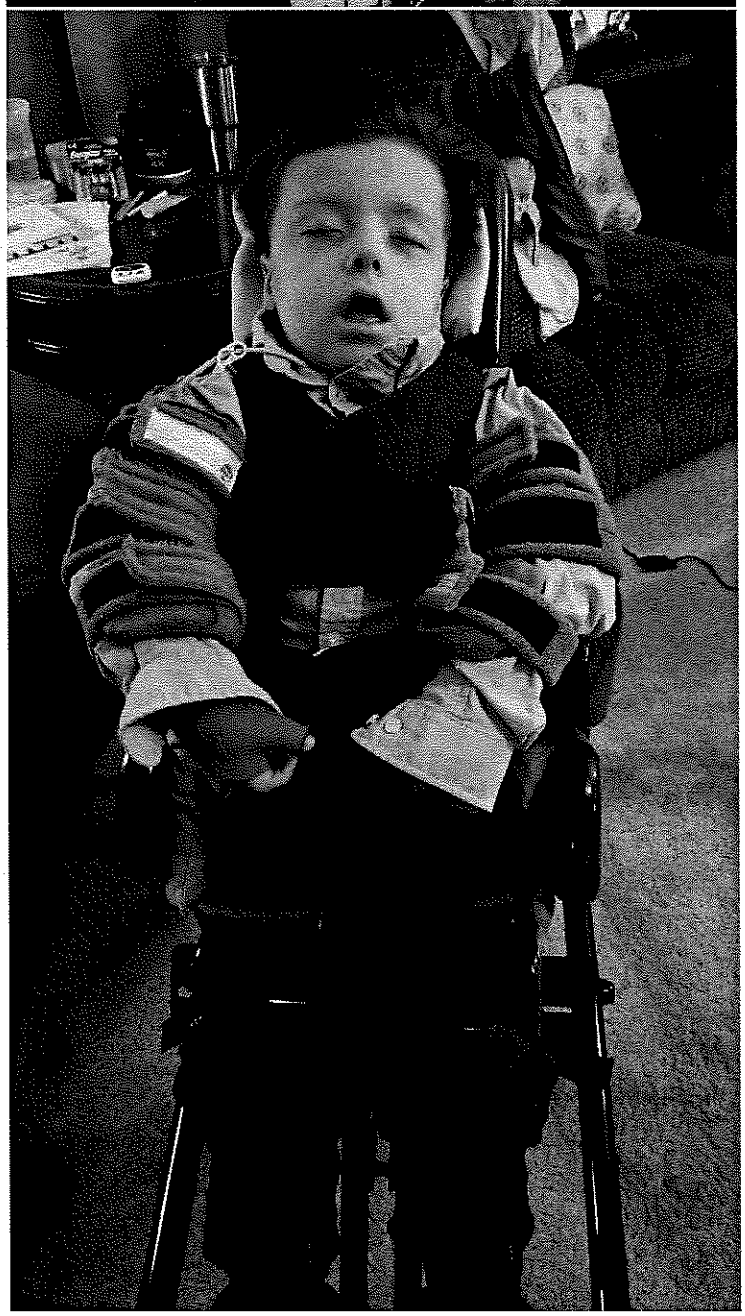
Whenever we traveled, we brought a van full of equipment. We needed to plan it very carefully to make sure he would be comfortable in his car seat. His schedule dictated when we would travel.

If Collin could have communicated with us, it would have helped us to provide him a better quality of life. Instead, it would take days for us to figure out what would help him. Life with Collin was hard on us all. Being his advocate, nurse, and parent – which came with his diagnosis – forced us into roles that we never thought possible.

We continue this fight because when a life, a child, a family is saved from the devastation of this disease, Collin's suffering will be providing hope to those important families. Senate Bill 194 need to be implemented to allow all Wisconsin babies a chance at life.

Sincerely,
Judy Cushman





15ml pediasure every 5 mins.
10ml water flushes

6 AM 4.4 ml Baclofen
7 AM 1.8 ml Gabapentin
8 AM 4½ oz VPS mix w/ 2 tsp Miralax
11 AM Vest/Neb (Albuterol + Budesonide)
Cough Assist 3-4-3
Wash/Brush Teeth
Eyedrops - 1 drop/eye - Warm Compress (microwave 8 seconds prior) Keep eyes closed 2 minutes

12 PM 3½ oz VPS + ½ oz H_2O
2 PM 4.4 ml Baclofen + 1 ml Multi-Vitamin
3 PM 1.8 ml Gabapentin
4 PM Vest / CA
4:30 PM Standard
5 PM 4 oz VPS
8 PM Vest/Neb/CA/Teeth/Eyes (Refer to 11 AM)
9 PM 4 oz VPS
10 PM 4.4 ml Baclofen 5ml Cetirizine .6ml Gas Relief
10:45 PM 9½ oz Pedialyte
11 PM 1.8 ml Gabapentin
.6 ml gas relief - PRN

School - Tues + Wed 12:15 Reading Buddies
Bath - Mon + Fri
OT - Thurs

Why Must Krabbe Disease Be Added to Wisconsin's Newborn Screening Panel?

Meet Jeremy. He's a 20-year old young man who likes Legos, X-box, YouTube, and movies. He also likes singing and puns. We know this because he's alive.

He's alive because he received a stem cell transplant when he was a month old. We knew he needed the transplant because we had him tested for Krabbe Disease when he was born. We knew to test him because his older brother Alex had the disease and died when he was 13 months old.

Currently, families who have the potential for Krabbe Disease only find out about it when they have a child, who, at birth, seems healthy and happy; his parents have plans and dreams for the future. Only a few weeks or months later, after noticing that "something is wrong" and after many, many doctor appointments, tests, monitoring, some mis-diagnoses, do they find out that their beloved child has Krabbe Disease. A disease that robs that loved child of the ability to swallow, move, hear, see, and eventually to live. A disease that robs his parents of that child's hugs, smiles, laughs, and future.

No child should ever have to endure the pain and suffering that occur with Krabbe Disease. The technology is available to test at birth. Other states have already adopted this technology and are saving children's lives. Wisconsin needs to be another one of those states. We, ourselves, know of at least 5 families in Wisconsin who have lost children from this horrible disease. They could have been treated and their lives saved had they known it was lurking behind the scenes.

Jeremy likes to quote movies. One of his favorite quotes (while it makes me shudder) is from Batman The Dark Knight movie.

"Do you want to know how I got these scars?" He says it because he's quoting Joker and to be funny. His dad and I *know* how he got those scars. He got those scars from the central lines he had during the stem cell transplant. We are HAPPY he has those scars. Those scars saved his life.

But.... He only has those scars because we knew to test him for Krabbe Disease. And it shouldn't take the death of an older sibling to know to do that.

Please add Krabbe Disease to the Wisconsin Newborn Screening Panel.

It's the difference between life and death. And as Jeremy likes to quote from one of his favorite Star Wars characters. "Do it!"

I want to thank the Wisconsin legislature for allowing me to briefly speak today in support of adding Krabbe disease to the state newborn screening panel. I am going to comment on the screen itself, estimated costs as well as performance data, all of which is published in peer-reviewed journals or available online from vendor catalogs. My lab at the University of Washington developed the Krabbe newborn screening assay that is used throughout the USA, and arguably I know more about this assay than just about anyone in the world today. I should also stay that the assay is in the public domain, neither the University of Washington nor myself have ownership of the assay, no patents, etc, and thus the remarks I make today have no bearing on financial benefits to myself or my employer.

The Krabbe newborn screening test is a two-stage process. The first stage is to measure the activity of the relevant enzyme in dried blood spots on newborn screening cards. Now that Wisconsin is screening for Pompe disease, the cost to add this additional enzyme assay for Krabbe is about $1-1.50 per newborn. This is the cost of the Krabbe-specific reagents. I am holding the catalog page in my hand, and the reagent is simply added to the same assay cocktail that is used for Pompe disease. Both enzymes are assayed together by the same process including the same disposable supplies, instrument for assay readout, etc, thus no additional supplies or personnel or equipment is needed beyond the $1-1.50 reagent. Several other states carry out the Pompe/Krabbe assay in this way, and this process has proven to work well. It is also published in a peer-reviewed journal.

Based on numbers in New York, where they have been screening for Krabbe disease for over a decade and on a total of about 3 million newborns, we can accurately predict about 50 newborns per year in Wisconsin will have a low Krabbe enzyme activity and are thus first-stage screen positive. Most of these will turn out to be false positives (not having the disease). To reduce the number of false positives, a second punch from the same dried blood spot is submitted to a second assay to measure a lipid called psychosine. The cheapest option for this is to send sample out to a second laboratory, for example PerkinElmer Genomics charges $99 per newborn. I have an email from the director of this lab with the price quote. So this adds about $5000 per year on top of the approximately $100,000 needed for the first stage reagents.

The Kentucky newborn screening lab has used this 2 stage process for the past 3 years and reports no false positives for Krabbe disease (I am holding the publication showing data for the first ~1 year).

During the past year, about 20 experts in the Krabbe field have met monthly to discuss guidelines for Krabbe disease newborn screening and follow-up; these guidelines will be published in the next few weeks. When the psychosine value is found to be above a critical value, the newborn is at very high risk for development of the most severe form of Krabbe disease (infantile form) and should be immediately transferred to a specialized center for evaluation for a bone marrow transplant. You will hear more about this in the testimony from Dr. Kurtzberg. If psychosine is found to be in the intermediate elevated range, the newborn is at very high risk to develop later onset Krabbe disease in the first decade of life. In these cases, a well-defined clinical follow up plan is described in the guidelines chapter.

Let's look at the numbers expected in Wisconsin based on the New York data. 0-2 severe

Krabbe patients per year and 3-5 high risk for late onset disease predicted per year in Wisconsin. All other families will be reported as screen negative. Note that this process is settled by the newborn screening lab prior to reporting out to the primary care physician and the families. So the number of families worried about Krabbe disease will be kept only to the most at-risk babies.

In summary, adding Krabbe disease to the Wisconsin panel will require < $2 per newborn for the test itself including all personnel, supplies and equipment as well as additional costs for clinical follow up on < 10 patients per year (most of which fall outside of the newborn screening lab). The false positive problem for newborn screening of Krabbe disease is no worse, and mostly better, than anything else now in newborn screening programs in the USA. Everything I have said today is in the peer-reviewed literature, is in vendor catalogs, and is supportive by ~20 of the world's top experts in Krabbe disease (who form the Krabbe disease consortium). Thank you for listening.

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State of Wisconsin
Department of Health Services
Tony Evers, Governor
Karen E. Timberlake, Secretary

TO: Members of the Senate Committee on Health

FROM: Dr. Robert Steiner, Newborn Screening Program Medical Consultant, Department of Health Services

DATE: March 17, 2021

RE: SB 194, relating to: newborn screening for Krabbe disease and requiring evaluation of additional lysosomal storage disorders for mandatory screening

Thank you for the opportunity to submit written testimony for information only on Senate Bill (SB) 194. SB 194 would require Krabbe Disease (KD) to be added to the list of conditions for which newborns are to be screened. It would also require the Department of Health Services (DHS) to evaluate on a biennial basis the appropriateness of mandatory newborn testing for lysosomal storage disorders in addition to Krabbe disease.

Newborn Screening has prevented death and disability for hundreds of Wisconsin residents since its inception in the mid-1960's. The Department of Health Services (DHS) oversees the state's Newborn Screening (NBS) Program and helps ensure success in screening, diagnosis, and treatment for Wisconsin newborns. The newborn screening currently tests for 49 disorders that are difficult to identify and are treatable if identified early. This includes testing for 47 blood disorders, a hearing screening, and screening for critical congenital heart disease. The NBS panel does not preclude families from seeking additional testing or genetic screens to identify a broader range of conditions. The program works very closely with the Wisconsin State Laboratory of Hygiene (WSLH), which is responsible for the blood screening laboratory testing.

Wisconsin has a formal process for evaluating proposals to add disorders to the NBS panel. The purpose of this process is to ensure that, before disorders are added, we can be sure that there will be an overall benefit to families and society, and to avoid negative or unintended impacts. It is a comprehensive, evidence-based approach which ensures that before a condition is added to the panel, there is an accurate test available, interventions are reasonably available, safe and effective, and the details of follow-up and management are delineated.

The process relies upon experts in the areas of: medicine and science; statistics and epidemiology; ethical, legal, social, and policy analysis; and laboratory medicine. It also includes representation from individuals with target conditions or their parents and incorporates testimony from patient advocacy groups.

There is a similar process in place at the federal level to evaluate conditions as to whether newborn screening is indicated. The Recommended Uniform Screening Panel is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening programs. Krabbe Disease is not currently on the Recommended Uniform Screening Panel and the most recent review of the disease by HHS did not change this recommendation.

To: Wisconsin Senate Committee on Health
From: Jeffrey W. Britton, MD, FAAP
Pediatrician, Sheboygan, Wisconsin

Re: Written testimony regarding SB194

March 16, 2021

Thank you for the opportunity to submit written testimony against Senate Bill 194, which would require all newborns in Wisconsin to be tested for Krabbe disease.

I currently serve on the DHS Secretary's Advisory Committee for Newborn Screening. This committee was created several years ago, under the Walker administration, for the purpose of carefully reviewing additions and deletions to the newborn screening panel. There was concern, at the time of the committee's creation, that a newborn screening test could be added without fully vetting all the issues related to screening for a particular disease.

Our committee reviewed a request for Krabbe disease screening in 2020. We did not proceed with a recommendation to start screening because some very import details still need to be worked out. The treatment for Krabbe disease detected by newborn screening is a bone marrow transplant in the first 1-2 weeks of life. Currently this treatment is not available in Wisconsin; in fact, it was (at the time of our meeting) only available at Duke University and Children's Hospital of Philadelphia. So, any baby detected by Krabbe screening would need to be immediately transferred to one of these centers. To date no process for making this immediate transfer has been built in our state. We would need agreements with medical transport aircraft, as well as assurance from insurance companies including Medicaid that such transfer and subsequent out-of-state care would be covered. Alternatively, a program for Krabbe treatment would need to be built at a Children's Hospital in Wisconsin. Because this process was not in place, we could not in good conscience advise the addition of this test to the panel.

As a general pediatrician I have, on many occasions throughout my career, been on the receiving end of a phone call from the State Lab of Hygiene telling me that a baby I care for has an abnormal test on newborn screen. When that happens it is essential that all the details about "what to do next" have been fully worked out, so that I can proceed as quickly as possible to get the baby the help needed. Legislation such as SB194 carries the risk of causing unexpected consequences, such as detection of a baby with a disorder without having the "what to do next" process worked out.

For these reasons I urge you to vote against this legislation, and to support the ongoing work of the Secretary's Advisory Committee as it carefully reviews all proposed addition and deletion requests for the Newborn Screening Panel.

I am happy to answer any questions you may have by phone (920-459-1454) or email (jeffrey.w.britton@aah.org).

WISCONSIN HOSPITAL ASSOCIATION

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March 17, 2021

To: **Members, Wisconsin Senate Committee on Health**

From: **Ann Zenk, Senior Vice President Workforce & Clinical Practice**
WHA Representative to the Wisconsin Newborn Screening Program Umbrella Committee

Subject: **WHA Opposition to Senate Bill 194 – Adding Krabbe to Newborn Screening Program**

A moving case can be made for many conditions that could be added to the Wisconsin Newborn Screening Program (NBS), like Krabbe disease, and WHA is sympathetic to those cases. WHA, however, believes it is because so many cases could be compelling that the NBS program must evaluate each proposed addition based on defined standards. If the condition meets the standards, the Wisconsin Department of Health Services (DHS) has the authority to promulgate administrative rules, subject to legislative review and oversight, to add the condition to the program's screening panel.

The physicians, nurses, dieticians, genetic counselors, parents, and other experts who support the NBS program evaluated Krabbe disease in 2016, applying the program criteria, and recommended against adding Krabbe disease to the NBS panel. Like other groups, including the Wisconsin Chapter of the American Academy of Pediatrics, WHA also does not support adding or deleting conditions through legislation rather than the established process. Any new information related to Krabbe can and should be presented to the committee in a renewed application.

While it may seem to legislators that the process only works when a condition is added, it's important for the committee to remember that the process works both when a condition is added and when it is not. As recently as March 2020, the newborn screening umbrella committee has recommended the addition of an inherited genetic condition called Pompe to the newborn screening panel and the DHS Secretary has subsequently moved forward on promulgating a rule to add the condition. By approving the condition, the committee of medical experts and ethicists found sufficient evidence that testing is accurate and treatment modalities are effective and accessible to approve conducting broad-based testing for the condition. The process has not stalled out, it has worked when a test has met specific criteria.

WHA is a supporter of much of the good work of the NBS program. The Wisconsin Statutes, in general, require every infant born in Wisconsin to be screened for certain congenital disorders that are identified in administrative rule. According to the DHS website, infants are currently screened for 46 disorders. The screening typically is accomplished through blood obtained from the newborn while in the hospital and sent on a blood collection card by the hospital to the Wisconsin State Laboratory of Hygiene (WSLH). Wisconsin hospitals purchase the blood collection cards from WSLH for $109 per card. DHS and WSLH use revenue from the cards to pay for the program, including lab costs, certain services for those identified with conditions such as subsidizing the cost of nutritional supplements, and other administrative costs. If one assumes about 64,000 births in Wisconsin each year, the card fees generate nearly $7 million in revenue annually for the program.

The fee for the blood collection card paid by hospitals is established in the NBS administrative rule chapter, DHS 115. DHS 115 also includes criteria for adding conditions to the NBS screening panel. On the NBS webpage, DHS lists its criteria for the evaluation of proposals to add conditions:

Criterion 1: Mandated testing should be limited to conditions that cause serious health risks in childhood that are unlikely to be detected and prevented in the absence of newborn screening.

Criterion 2: For each condition, there should be information about the incidence, morbidity and mortality, and the natural history of the disorder.

Criterion 3: Conditions identified by newborn screening should be linked with interventions that have been shown in well-designed studies to be safe and effective in preventing serious health consequences.

Criterion 4: The interventions should be reasonably available to affected newborns.

Criterion 5: Appropriate follow-up should be available for newborns that have a false positive newborn screen.

Criterion 6: The characteristics of mandated tests in the newborn population should be known, including specificity, sensitivity, and predictive value.

Criterion 7: If a new sample collection system is needed to add a disorder, reliability and timeliness of sample collection must be demonstrated.

Criterion 8: Before a test is added to the panel, the details of reporting, follow-up, and management must be completely delineated, including development of standard instructions, identification of consultants, and identification of appropriate referral centers throughout the state/region.

Criterion 9: Recommendations and decisions should include consideration of the costs of the screening test, confirmatory testing, accompanying treatment, counseling, and the consequences of false positives. The mechanism of funding those costs should be identified. Expertise in economic factors should be available to those responsible for recommendations and decisions.

The criteria recognize that there is a cost when new tests are added to the program. For the addition of a test for Krabbe disease, it has been estimated that the cost of the blood collection card would increase by $4. For the addition of Pompe disease, another condition currently under consideration, DHS has estimated an increased cost of $10 for each card. If both diseases are added to the program, the annual cost of the program might increase by over $1,000,000 per year. With hospitals already bearing the burden of funding non-hospital services, such as the nutrition support funded by newborn screening card fees, funding sources must be identified before unbudgeted costs can be added to the NBS program.

As is the case for many worthwhile programs, it can be difficult to discuss program costs and funding sources. Funding for the program is not a new issue and we believe legislators should be part of the discussion. WHA believes the NBS program is an important public health program that, as such, should be funded with public dollars, rather than fees on our state's hospitals, and subject to the oversight expected of public programs.

KrabbeConnect
CONNECTING PATIENTS & RESEARCH

March 15, 2021

State Senator-Patrick Testin
Staffer-Jeff Schultz

Re: Collin's Cushman's Law

I sincerely regret I am unable to join in-person to give this testimony. I am a rare disease mom with more than 15 years of experience in the rare disease space. I have been blessed to experience many roles in the rare disease space with specialty pharmacy, pharma, philanthropy, but most importantly as a caregiver. I currently hold the position of President at KrabbeConnect, a non-profit organization working hard to reach a day in which each patient receives an early diagnosis, has access to state-of-the-art care, and lives a life free of Krabbe disease.

Krabbe disease is described as a severe neurological condition that results from the loss of the myelin sheath surrounding nerve cells. This protective myelin sheath is essential to insulate the nerves and ensure the rapid transmission of nerve signals throughout the body. When the myelin sheath around the nerves is worn away, damaged, or deteriorates, it causes problems throughout the body. Patients impacted with the infantile form of Krabbe disease experience the inability to swallow, severe irritability (above and beyond colic), loss of milestones (unable to hold their head, sit, crawl), seizures, and so much more.

It appears our society becomes overly focused on numbers and averages. The more people impacted, the larger the impact and or reward to our country, community, and families. I will be honest; this disease affects approximately 40 patients worldwide per year. At least, that is what the experts believe to be the case today. In the United States, the formal statistic for Krabbe disease has been reported as 1 in 100,000 individuals.

I strongly believe in the bill to add Krabbe disease to Wisconsin's newborn screening panel for the following reasons:

I. A suitable testing protocol is in place. Today, newborns can be initially screened by measuring GALC enzyme activity. If the enzyme activity is found to be low, a second-tier test will be utilized to look for the biomarker known as psychosine. The Mayo Clinic (Rochester, MN), under the direction of Dr. Dietrich Matern, has developed easy-to-use charts to help laboratories navigate Krabbe disease newborn screening. Also, Mayo clinic performs

second-tier testing for states interested in outsourcing to confirm and/or deny a Krabbe disease diagnosis.

- Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase
- Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase and Psychosine
- Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase, Psychosine, and GALC 30kb Deletion

II. Hematopoietic Stem Cell Transplant is a treatment proven to provide a patient a better quality of life if administered early on, preferably before 30 days of life.

- Studies show a strong improvement in lifespan and quality of life compared to patients not transplanted.
- Outcomes are more favorable when infants are transplanted before the first month of life.

III. Krabbe disease resource maps are available to help states, medical staff, and families quickly navigate their options for treatment once diagnosed.

IV. Gene therapy clinical trials are another option for treatment. These clinical trials require patients to be identified in the first few weeks of life. Today, 2 companies have committed to bringing a new treatment to the bedside of patients and they are:

- Forge Biologics-enrollment began this month for their combination therapy. Patients undergo a transplant and then receive gene therapy shortly after.
- Passage Bio-approved gene therapy only clinical trial but not enrolling as of this March 15th, 2021.

Many families in Wisconsin have lost loved ones to this disease. As I take a moment to reflect, I can name 7 Wisconsin patients that have died in the past 10 years from Krabbe disease. Most of them did not have the opportunity to celebrate their second birthday. Future Krabbe families deserve a fighting chance against this devasting disease. Please grant that to them. Believe me, the next family in Wisconsin will be forever grateful. Just ask any of the families from Kentucky, Ohio, Missouri, Indiana, Tennessee, Pennsylvania, and New York. These states have added Krabbe disease to their newborn screening panel and the Krabbe disease patients are thriving.

In closing, I want to personally thank the Cushman family and Senator Testin for their work and commitment to ensure the health and welfare of those living in the state of Wisconsin includes rare diseases such as Krabbe.

Warm Regards-

Stacy L. Pike-Langenfeld
President and Co-Founder, KrabbeConnect

Hunter's Hope Foundation

Krabbe ~ Leukodystrophies ~ Newborn Screening

March 16, 2021

Senator Patrick Testin
Room 8 South
State Capitol
PO Box 7882
Madison, WI 53707

RE: SB 194

Dear Senator Testin and the Distinguished WI Senate Committee on Health Members:

On behalf of the Hunter's Hope Foundation, I am writing to request your full support of Collin Cushman's Law to add Krabbe Disease to the Wisconsin Newborn Screening panel.

Hunter's Hope was established in 1998 by NFL Hall of Fame Quarterback Jim Kelly and his wife Jill when their son Hunter (2/14/97-8/5/05) was diagnosed with Krabbe Leukodystrophy. If left untreated, Krabbe is a devastating and fatal disease. The only available treatment must be administered prior to the onset of symptoms, making newborn screening (NBS) for Krabbe *crucial* to giving affected children a fair chance at life.

For nearly twenty years Hunter's Hope has partnered with the world's leading disease and NBS experts to make newborn screening for Krabbe a successful and viable option for every state. The experts agree that Krabbe NBS is ready for widespread implementation and are moving forward with efforts to have Krabbe added to the Recommended Uniform Screening Panel or RUSP. However, each state still must determine what diseases to screen for and it can take years to fully implement new disorders. Tragically, any delay in implementing Krabbe NBS in WI can lead to additional children in this state needlessly suffering and dying from a treatable disease.

The Hunter's Hope Foundation has had the privilege to work alongside the Cushman family over the past several years as they have tirelessly advocated for Krabbe newborn screening to be added in Wisconsin. The Cushmans have followed the tedious path laid out by the state's health department to no avail.

We urge the members of this committee to make a stand for future WI children to have the opportunity for lifesaving treatment by mandating the addition of Krabbe Disease to the state's newborn screening panel.

Hunter's Hope Foundation

Krabbe ~ Leukodystrophies ~ Newborn Screening

Hunter's Hope has a system in place to support states with Krabbe NBS through our Annual Scientific and Medical Symposium, and our Krabbe NBS Council which has monthly webinars. These efforts gather all stakeholders and experts to ensure the best possible outcomes for children identified through Krabbe NBS.

If New York, Missouri, Kentucky, Ohio, Tennessee, Illinois, New Jersey, and Indiana can successfully screen for Krabbe at birth, so can Wisconsin.

Please support this effort and save future children like Collin…

With hope,
Anna Grantham

Anna Grantham
NBS Director
Hunter's Hope Foundation

2021 Senate Bill 194
Newborn Screening for Krabbe Disease
Testimony of Norman Fost MD MPH

Submitted in writing to Senate Committee on Health Clerk, Heather Smith, Heather.Smith@legis.wisconsin.gov

Copy to Sen. Patrick Testin, Sen.Testin@legis.wo.gov
Copy to Sen. Kelda Roys, Sen.Roys@legis.wisconsin.gov

I am Professor Emeritus of Pediatrics and Medical History and Bioethics at the University of Wisconsin School of Medicine and Public Health, and Chair of the Wisconsin DHS Secretary's Advisory Committee on Newborn Screening. I have served on numerous federal committees related to newborn screening since 1975 and authored numerous scholarly articles on ethical, legal and policy issues in newborn screening.

Newborn screening has saved innumerable lives and prevented serious disability in millions of children world wide. It is one of the great advances in public health in the past 100 years, and Wisconsin has been a leader in these advances.

Premature introduction of newborn screening, without adequate review by experts in science, ethics, and law has killed or caused severe brain damage in thousands of infants in Wisconsin and throughout the United States. Examples are provided in the attached article (pp 242-251). It has also led to widespread confusion among parents due to false positive tests and major reproductive decisions based on misinformation. Wisconsin has also been a leader in some of these problems.

Because of these problems, in 2013 I was asked by the Walker Administration to re-organize Wisconsin's process for adding tests or disorders to the newborn screening program. I did so, and Wisconsin now has a model process for review of proposals to add tests to the screening program, including leading experts in relevant disciplines, representatives from hospitals and the physician community, parents of children with disabilities, and involvement by the public. We work in collaboration with a highly respected US DHHS advisory committee on newborn screening.

Screening for Krabbe Disease has been extensively reviewed by our Committee twice in the last five years, most recently in November 2020 (see attached report to Secretary Designee Andrea Palm). We concluded that screening for Krabbe Disease at this time would result in serious harm to some children, and uncertain benefits. We look forward to reviewing continuing advances in this field, and re-visiting the proposal as new information becomes available.

With regard to the specific language of SB 194, I have several specific questions, among them:

- How will an attending physician know where or how to obtain the test?
- How much will the test cost?

- Who will pay for it?
- Will parents be given the opportunity to consent to this controversial test? Who will inform them of the relevant information? Will the physician who is required to obtain the test know the relevant facts? How much time will it take to review this information with parents so they can make an informed decision about whether to consent to the test?
- If the physician learns his/her patient has a positive test, what is s/he supposed to do then? Who should s/he contact?
- If an infant has a positive test, is effective treatment available? Where? What is the evidence of the safety and efficacy of that treatment? Who will review that information with the parent(s)? How much will the treatment cost? Who will pay for it? How soon must the treatment be started? If only available out of state, who will arrange for transportation and lodging? How long will the family need to reside out of state? Who will pay for that?

In addition to these specific questions, I am very concerned that adding Krabbe screening through the legislative process will cause permanent damage to Wisconsin's outstanding review process, as concerned parents will correctly foresee that they can find the response they want through the legislature. I am certain this will result in substantial harm to many children and families, with uncertain benefits.

B.Strong for Bryce
Kyle & Jenna Heckendorf
805 Erik Street
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Senate Bill 194: Relating to: newborn screening for Krabbe disease and requiring evaluation of additional lysosomal storage disorders for mandatory screening
Submitted Written Testimony of Kyle & Jenna Heckendorf
Senate Committee on Health
March 17, 2021

Thank you, Chairman Testin, Ranking Member Erpenbach and committee members, for your time in holding a public hearing and taking testimony on this bill as it is very personal to us because we lost our first child, Bryce Thomas, to Krabbe disease at just 18 months old. Thank you for giving us this opportunity to tell you about our son and the importance of including Krabbe Leukodystrophy on the mandatory newborn screening panel here in Wisconsin.

On May 6th, 2013, we welcomed Bryce into the world. Life was perfect. Our beautiful, healthy little boy was here and as first time parents, we were already looking forward to all of life's special moments ahead. Bryce was a very content and happy baby. We adored his smiles, giggles, play time and watching him develop his little personality. Developmentally, Bryce met every milestone for the first five months. At around five and a half months, however, we started noticing some changes: his neck seemed weaker, he absolutely hated tummy time, his body seemed more stiff and rigid, his appetite decreased, and it seemed as if he didn't move his arms and legs as much. These beginning symptoms continued to become more alarming and we knew something wasn't right. For the next couple weeks, Bryce was in and out of clinics and hospitals in Dodgeville and Madison as doctors tried to figure out what was wrong. Bryce underwent all kinds of testing, and we continued to hold onto hope as doctors ruled out many different possibilities. On November 19th, 2013, a blood test finally confirmed Bryce had Krabbe disease and he would probably not live to see his second birthday. Our world was flipped upside down. Everything we were looking forward to — hearing Bryce say his first words, watching him take his first steps, helping him catch his first fish, playing catch in the backyard, simply watching him grow up — were not going to happen and that news was absolutely devastating.

"Your son is going to die." These are the most heartbreaking words any parent can hear. The doctors and genetic counselors offered no hope. They said it was too late to do anything. Once symptoms of Krabbe disease are present, it's too late. There was no way to prevent Bryce from dying. No treatment was offered that might slow the disease from

progressing. Doctors told us that the only thing they could do for Bryce was to keep him comfortable as he would slowly lose his ability to move his arms and legs, smile, eat, swallow, see, hear, and eventually breathe. Bryce's battle with Krabbe was anything but comfortable. He screamed in pain for hours on end as the disease progressed and his nerves became exposed. He vomited after nearly every meal until surgery was required to prevent this from happening. He clenched his hands so tight that we had to put sponges in them to offer some sort of relief. He endured many hospital stays and surgeries. When Bryce no longer could swallow, he required suctioning around the clock. Near the end of his life, he was hooked up to an oxygen machine continuously. Bryce lost his torturous battle with Krabbe at 18 months old on November 26, 2014.

By putting Krabbe Leukodystrophy on the mandatory newborn screening panel, no parent in Wisconsin, from here on out, will be left without hope. Detecting this horrific disease at birth is essential and will give every child born with Krabbe a fighting chance. *Every* child deserves that chance — the chance that our amazing little boy did not get.

Through our journey with Krabbe disease, we have met many amazing families and have seen first hand the hope a transplant can bring. For example, Michael Wilson, from Oregon, was diagnosed at birth with Krabbe. He received a transplant and is now a thriving 10 year old with no signs of the disease. For Michael and many others, Krabbe was not a death sentence. Those children and their families are so fortunate to have been given a fighting chance. We would have given anything to have had that chance with Bryce. Krabbe will remain a death sentence for every child born in Wisconsin with this disease if it is not screened for at birth. You, as a committee, have the opportunity to save future babies born in our state and give hope to their families.

We'd like to thank everyone who has put so much time and effort into the pursuit of giving children in Wisconsin born with Krabbe Leukodystrophy a chance to receive a life-saving treatment. Thank you for reading our story and please reach out if you have any questions (jheckendorf@rvschools.org). We would greatly appreciate your support of Senate Bill 194.

Sincerely,

Kyle Heckendorf

Jenna Heckendorf

Kyle & Jenna Heckendorf