MD Krabbe bill SB0117 written testimony.docx.pdf
Uploaded by: Brian Stone
Position: FAV

15 February 2024

The Honorable Pamela Beidle
Chair, Senate Finance Committee
3 East Miller, Senate Office Building
Annapolis, MD 21401

Subject – SB0117- Health - Newborn Screening Program - Krabbe Leukodystrophy

My name is Brian Stone and I am the uncle of Lillian Grace Smith. You will read and hear testimonies from families impacted by Krabbe and how not being screened at time of birth has impacted their child's and the entire family's lives and the importance to pass this newborn screening bill for Krabbe. What I wanted to walk each of you thru is what a typical day is like for Lily, age 12, diagnosed at 6 months, transplanted at 7 months.

7:00 am – nebulizer treatment, shaky vest, cough assist, 2nd nebulizer treatment, trachea care followed by medications and flush – lasts about 45 minutes

9:00 am – food, medications and flush, bath, change of clothes, physical therapy for 1.5 - 2 hours

11:00 am – food and flush, back to bed for side laying, position change

12:00 pm – occupational therapy and/or speech therapy

1:00 pm – food, medications and flush

3:00 pm – food, medications and flush, change of clothes, positional change to lay on couch, medications

5:00 pm – food and flush, positional change

7:00 pm – food, medications and flush, nebulizer treatment, shaky vest, cough assist, 2nd nebulizer treatment, trachea care

9:00 pm – food, medications, and flush, get ready for bed.

REPEAT. Day after day. 7 days a week. 52 weeks a year.

Note - Every hour someone needs to change Lily's positions to avoid bed sores.

A schedule of other therapies IF its during the school year, IF therapists/teacher are available and IF Lily and therapist/teacher are not sick

Monday – Physical therapy for 45 minutes for head control
Tuesday – home teacher for 1 hour
Wednesday – School for occupational therapy and physical therapy
Thursday – Private Occupational therapy
Friday – virtual speech therapy

All of that care is needed in a typical day, yet Lily is the bravest and strongest person I know and is one of Krabbe's biggest transplant success stories. Most of this could have been prevented if the state of

Maryland had screened Lily for Krabbe at child birth. Lily would have been diagnosed early on, received a successful transplant and had a very normal day to day life as a 12 year old. If costs are concerned, let me assure you that the cost to care for Lily and other Krabbe children in the state of Maryland FAR exceeds any costs to acquire an additional testing unit and around the clock coverage to man perform screening. That's not including the emotional toll it takes on a Krabbe baby's family, friends and community. Give Maryland families the knowledge of what their child faces and give them the opportunity to make a decision on what is best for their family immediately after child birth. Thank you for your time and looking forward to helping pass this important bill along with you.

Sincerely,

Brian Stone
23109 Hayden Court
Lexington Park, MD 20653

Krabbe testimony-SB0117.pdf
Uploaded by: Jessica Blackwell
Position: FAV

15 February 2024

The Honorable Pamela Beidle
Chair, Senate Finance Committee
3 East Miller Senate Building
Annapolis, MD 21401

Subject: **SB0117 – Health- Newborn Screening Program - Krabbe Leukodystrophy**

Chair Beidle, Vice Chair Klausmeier and Members of the Finance Committee:

My name is Jessica Blackwell and I am Lily Smith's aunt; her mom is my sister Kathleen. I am writing this with the intent to support the passing of a law that would add Krabbe testing to the newborn screening that is performed in the state of Maryland. Lily is our family's little angel. She was a happy and healthy baby until she was 6 months old and started exhibiting symptoms of Krabbe. We did not know what it was at the time but her momma knew something was not right and they received a diagnosis from Children's National Hospital. The doctors there told her parents to take her home and enjoy the time they had left while making her comfortable because she would not live past her first birthday. Her parents were fighters and found a very specialized transplant protocol through Children's Hospital of Pittsburgh that would hopefully stop the progression of the disease. While this transplant did stop the progression of the disease, she would never regain what had been lost during those first six months when no one knew she had Krabbe because it is not part of the newborn testing in Maryland.

Because of the lack of testing in Maryland, Lily is wheelchair bound with a feeding tube and a trachea and requires constant care. Because there is no testing in Maryland Lily has celebrated her first, tenth and twelfth birthdays in hospitals rather than at Chuck E. Cheese playing with friends. Because there is no testing in Maryland, Lily cannot ride a bike or run outside like other 12 year old children. Because there is no testing in Maryland, Lily cannot attend school and play during recess; instead she works harder than any person I know to be able to hold her head up and try to communicate through an eye gaze device, the only part of Lily's body that she has control over. If Lily had been born in a state that included Krabbe as part of their newborn screening, she would have a completely different life. If Krabbe had been included in Maryland's newborn screening when Lily was born, she could have had the transplant that stopped the progression of the disease right after birth and she would be running around and riding a bicycle like other young children.

I do not understand why Maryland has not added Krabbe to newborn screening. I came here approximately ten years ago asking for the same thing – add Krabbe to newborn screening in the state of Maryland - and still nothing has been done. If adding this screening can save even one child and one family from experiencing the pain and hardships that Lily and her family experience on a daily basis then I do not understand how the state of Maryland can vote against including this testing as part of newborn screening.

Respectfully,

Jessica A. Blackwell

SB117 - Smith.pdf
Uploaded by: Kathleen Smith
Position: FAV

Feb 15, 2024

The Honorable Pamela Beidle
Chair, Senate Finance Committee
3 East Miller Senate Office Building
Annapolis, Maryland 21401

RE: SB117- Health – Newborn Screening Program – Krabbe Leukodystrophy

Dear Chair Beidle, Vice-Chair Klausmeler and Members of the Finance Committee:

My name is Kathleen Smith, and I am a mother of three beautiful children. My youngest daughter came home from the hospital as a healthy baby, and we had all the dreams a new parent has for their child. At five months old Lily began regressing in her milestones. She was no longer able to hold her head up or grab for toys like she had been able to just days before. She began crying a high-pitched cry I had never heard before and stretch her back in pain. My baby was no longer healthy, and I did not know what was wrong. We headed to the pediatrician who agreed that she was declining and recommended that she be seen by a developmental pediatrician. I called every developmental specialist I could get ahold of, and they all had a several month waiting period and I just could not stand seeing my daughter in such pain. We took her to Children's National ER where the doctors took her right back – I showed them videos of Lily playing with toys in a boppy seat like any other child, which was just days ago. Now Lily could not even hold up her head on her own, she was very stiff, she kept her thumbs always squeezed in her hands and she had a cry no parent should ever hear. They rushed her back for a CT scan, I'll never forget them telling me she had white matter on her brain. Like any parent, I felt guilty, was this something I had done, I had not provided enough nutrients when I was carrying her, had I bumped her head and not even known. What had I done to make my baby cry this horrible cry and be in such pain. They assured me it was nothing I had done, but I still did not believe them – I am a Mom, I was responsible for this perfect baby. They wanted to admit her as a patient for a sedated MRI – the next morning she had the sedated MRI, the results were not good. It seemed like forever; I am certain it was only hours before someone came and asked if Ben (my husband) and I were related – we insisted that we were not, and they asked lots more questions that just seemed odd – finally we said what is WRONG with our baby??? They looked down at the ground and said that they were 99% sure that she had Krabbe Disease and that there is nothing that can be done expect to keep her as comfortable as possible and she would not live to see her second birthday. I remember falling on my knees in complete disbelief that my daughter was going to die a very painful death. It was at this time that we asked in disbelief, there is nothing that can be done? They said IF we had caught it earlier, she could have received a stem cell transplant, but that since she is

already symptomatic there is nothing that can be done. They said to take her home, contact Hospice and take lots of pictures she would not live to see her 2nd birthday.

We have family in the medical profession, and they researched and found NIH studies for Krabbe and that is how we saved Lily's life. We took her to Pittsburgh to meet a very special doctor who has spent her life researching Krabbe and learning as much as she can about helping these children. She performed lots of tests on Lily and gave us the option to have Lily transplanted even though she was symptomatic. Of course, we did not want to keep Lily in pain, so we asked what her life would be life after the transplant, the assured us that she would keep her vision, hearing and cognitive abilities. Lily can't walk, can't speak, BUT Lily is HERE with us!! She CAN SMILE and make others SMILE. Lily is amazing, she uses an eye gaze device to speak, play games, and read books. She loves to be with her family, go outside for walks, go camping, blow bubbles with her switch toys and of course watch her shows.

If Lily had been caught through newborn screening, you would not be able to tell that Lily ever had Krabbe. We know families who have lost a child to Krabbe, tested a sibling at birth and received the transplant prior to symptoms and these children are completely normal – no one would ever know that they had Krabbe. This disease progresses very quickly, there is no time to waste. The only way to save children from Krabbe is to test them as newborns and treat them prior to symptoms. We were lucky to get diagnosed as quickly as we did, if we had waited to see the developmental specialist, it would have been too late and Lily would not be here today. Unfortunately, many doctors do not know about Krabbe and often misdiagnose children as having colic, cerebral palsy, or many other illnesses. Many families go through a diagnostic odyssey before finding out it is Krabbe Disease and all the while the child is losing abilities, the parents are losing a child (not even aware) and the child is in pain from not being on the correct medications to keep them comfortable.

Please stand up for these children and add Krabbe to newborn screening in Maryland!

Sincerely,
Kathleen Smith
Mom of Lily
240-538-3077

Shoemaker Family Testimony 2.15.24.pdf
Uploaded by: Melissa Shoemaker
Position: FAV

February 15, 2024

Shoemaker Family
1811 Hatfield Rd
Huntingtown, MD 20639

Senate Bill 117
Finance Committee
East Miller Senate Building, Room 3
Annapolis, MD

Dear Senate and Finance Committee Members,

My name is Melissa Shoemaker, our family has been forever changed by the choice the state of Maryland has made to not screen newborns for Krabbe Leukodystrophy at birth. My son Parker Eugene was robbed at his opportunity for lifesaving treatment by being born in Maryland versus another state that screens for Krabbe Leukodystrophy at birth as a standard practice. Although not a curable disease, there is treatment for Krabbe especially if diagnosed early, before becoming symptomatic through Newborn Screening. My family was not given the choice to treat him as he was already affected by the disease and presented symptoms at the time of diagnosis. I have met children that were born with the same disease as my son, who are breathing, talking, laughing, walking, and living all the wonders of an ordinary normal wonderful life. It is truly heartbreaking to know what we now know about life saving early detection.

When asked to testify I was both honored and fearful. Our family has attempted and failed on many occasions to persuade the state to take into consideration all the research, facts, and real-life witnesses. Reliving our life with Parker is so beautiful and yet so very painful. Our story will hopefully help someone in this room, and that is what is most important to us and Parker's legacy. Knowing that this bill could save a family the pain and suffering of the wisdom discovered too late, the months of not knowing what is wrong with their newborn, and the inevitable deterioration and loss of their child. Knowledge is power and we strongly advocate for the education of diagnosis for all parents in Maryland.

Our first child, Parker Eugene was born on 11/9/2014 at Anne Arundel Medical Center in Annapolis, MD. My husband and I had already been married for four years and together for ten before welcoming our sweet, red-haired baby boy, and we were so excited to start our family. We were filled with hopes and dreams for the future and all the things' parents get to enjoy with their children as they grow.

The first few months were exhausting, and joyous. They were amazingly hard, and also the happiest days of our lives. Parker was fussy, and cried often but we were reassured that he was just fine, and this was typical behavior for a colicky baby. He had difficulties feeding and seemed to always have gas or an upset stomach, again reassured that this is also normal for a newborn with an immature GI system. He had failure to thrive/gain weight. He was diagnosed with reflux and milk intolerance and prescribed medicine and special formula. I became truly concerned right around 4 months. He would cry during feedings, he did not try to reach for his toys on his mat as he once did, he could not sit up as well, he had jitters in his right hand, he was in so much pain, and he could not sleep. I took him for his 4-month appointment and expressed all my concerns, I read them from a list. The doctor was an old, trusted friend, and he was not

worried. He reassured me that Parker was okay. I insisted on seeing a neurologist, he complied and gave me a number to call. The next day I called and was advised that the only way to get an appointment was to be the first in line on the day when you call. I called every day at 8:00am for four days in a row and never was given an appointment. A week later, my MIL was concerned he had not been eating all day. I left work and took him to the doctor and was sent to the ER with fear that he was showing symptoms of CP due to scissoring in his legs and fear of dehydration. It all happened so fast. After a CT scan, and blood work, they did not find anything to be abnormal and sent us to Children's via ambulance. We checked into the 4th floor with no trouble as a team was waiting for us. It was overwhelming to see all the staff and hearing the words metabolic tossed around. I had no clue what that meant and assumed it was a good thing, an easy fix. I remember taking a photo of the room number so I could show Parker when he was older and tell him about that crazy first time in the hospital. He would want all the details and to hear about how brave he was. My husband and I were scared, and hungry, we had no clothes or toiletries. We wanted to know what was wrong and what the plan was to get our boy to feel better. The number of doctors and nurses in and out of our room was overwhelming but assumed normal protocol.

Parker received his first MRI at Children's National on March 25th, 2015, he was 4 1/2 months old. That evening the genetics team came to tell us their initial diagnosis. They were pretty confident that Parker had Krabbe Disease. My first thoughts were "this sounds manageable," I had never heard of it so how could it be that bad? Their faces seemed so solemn, and they then shared the life expectancy which was 2 years. Little did we know we would not even get one. The next few hours were pure life shattering, heartbreaking, soul crushing devastation. We stood over Parkers crib and sobbed. On Friday March 27 the blood test confirmed that our beautiful son had Krabbe Leukodystrophy. That night I wrote this: "My baby will never grow up. He will never talk or walk. He will never play sports or ride a bike. He will never go to prom or college. My baby has Krabbe Disease. Today we found out our sweet boy has a fatal illness. Nothing can describe my pain and sorrow. I will bury my only child before his second birthday. My life, Gene's life, Parkers life have been taken from me. The prayers did not work."

We were referred to the Children's Hospital of Pittsburg to see Dr. Escolar who had performed cord blood transplants, the only treatment available for Krabbe disease, and she was the nation's leader in Krabbe research. We were discharged that Friday and headed up to PA on Sunday. We were holding on to the hope that Parker could be a candidate for transplant. After countless tests and evaluations, it was decided that Parker was not eligible for treatment, because he was already symptomatic the treatment would not be successful. He received a G-Tube to help with feeding and we were sent on our way to help our baby slowly transition into the arms of the lord in heaven.

So much love, pain and suffering occurred during diagnosis day to the day my son passed away in my arms at 10 months old on September 14, 2015. I am leaving out all the details of palliative care and prayers for a miracle. All the medications and the progressive loss of ability including swallowing, coughing, general movement. The machines needed to keep him comfortable and alive. The search for second opinions.

My ask today is that the committee take into consideration the addition of Krabbe to the NBS panel knowing there is life-saving treatment available, that the capabilities and science are already available to the state and the recent decision from the federal advisory board to add Krabbe to the RUSP. (Recommended Uniform Screening Panel)

Knowledge is power and we strongly advocate for parents in Maryland to have the education of early diagnosis.

Our families right to decide if treatment was a good fit for our son was taken from us. Our diagnosis came too late, but we believe that the right to choose treatment should be given to every family to decide on their own. We are advocating for SB117 to have a favorable outcome. The capabilities and science are already available, and treatment is proven to be successful under the right circumstances including early detection.

Thank you for your time and careful consideration.

Parker's Mama
Melissa Shoemaker
443-871-4294
Melharris001@yahoo.com

SB0117-543026-1.pdf
Uploaded by: Jack Bailey
Position: FWA

SB0117/543026/1

AMENDMENTS PREPARED BY THE DEPT. OF LEGISLATIVE SERVICES

13 FEB 24
15:55:34

BY: Senator Bailey
(To be offered in the Finance Committee)

AMENDMENTS TO SENATE BILL 117
(First Reading File Bill)

AMENDMENT NO. 1

On page 1, in line 2, after "**Program** –" insert "**Infantile**"; in the same line, strike "**Leukodystrophy**" and substitute "**Disease**"; in line 4, after "for" insert "infantile"; and in the same line, strike "leukodystrophy" and substitute "disease".

AMENDMENT NO. 2

On page 2, in line 4, after "**FOR**" insert "**INFANTILE**"; and in the same line, strike "**LEUKODYSTROPHY**" and substitute "**DISEASE**".

SenatorBailey_FWA_SB117.pdf
Uploaded by: Jack Bailey
Position: FWA

JACK BAILEY
Legislative District 29
Calvert and St. Mary's Counties

Budget & Taxation Committee

THE SENATE OF MARYLAND
ANNAPOLIS, MARYLAND 21401

Annapolis Office
James Senate Office Building
11 Bladen Street, Room 401
Annapolis, Maryland 21401
410-841-3673 · 301-858-3673
800-492-7122 Ext. 3673
Jack.Bailey@senate.state.md.us

District Office
Dorsey Professional Park
23680 Three Notch Road, Unit 101
Hollywood, Maryland 20636
240-309-4238

February 16, 2024

Senate Bill 117 – Health - Newborn Screening Program - Krabbe Leukodystrophy

Dear Chair Beidle and Members of the Committee,

I am writing to introduce Senate Bill 117. This bill would require that Krabbe Disease be added to the Maryland Department of Health's newborn screening system.

Krabbe is a neurological disease caused by the absence of an enzyme which leads to the loss of myelin causing the nerves and brain to be unable to communicate. Symptoms begin mildly around 4-6 months of age but quickly progress to blindness, deafness, seizures, decreased mobility and eventually death. If detected at the beginning of life, Krabbe is treatable. Early detection of Infantile Krabbe Disease allows for a transplant which can dramatically improve the quality and longevity of the child's life. However, this operation can usually only be done before the child is six months of age. Far too often, by the time the child is diagnosed, it is too late for a transplant. Adding Krabbe to the newborn screening system will save the lives of children in our State.

Last month, the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended Krabbe be added to the Recommended Uniform Screening Panel for newborns. The Secretary of Health and Human Services will make the final determination, but Senate Bill 117 will allow Maryland to move forward without delay with this life-saving screening for newborns. Unfortunately, we have seen our State react slowly to the recommendations of the federal government in adding to our newborn screening system. In preparing to present this legislation, I was made aware of two other diseases that have been recommended by the ACHDNC but not added by Maryland's advisory committee, which is responsible for advising the Secretary of Health, who has the final authority to add these diseases to the newborn screening system in this State.

Eleven other states have instituted this screening. Nine of these eleven states have done so since the General Assembly last considered adding Krabbe to the screening system in 2014. These states did so before the federal advisory committee made their recommendations. We have more backing with regards to the scientific evidence than any of these other states had when they made their decision. It is time to add Infantile Krabbe Disease to the list newborn screening system.

I am submitting with this bill an amendment to specify that the bill applies to Infantile Krabbe Disease, to be consistent with the Federal recommendation. I respectfully request a favorable report with this amendment on Senate Bill 117. Thank you for your consideration.

Sincerely,

Senator Jack Bailey

Hamosh_SB117_opposed.pdf
Uploaded by: Ada Hamosh
Position: UNF

Ada Hamosh, MD, MPH
1104 Ryegate Road
Towson, MD 21286
Email: ahamosh@jhmi.edu

February 13, 2024

Senator Pamela G. Beidle
Miller Senate Office Building
3 East Wing
11 Bladen St.
Annapolis, Maryland 21401

Re: SB117/HB095 Health - Newborn Screening Program – Krabbe Leukodystrophy
POSITION: Oppose

To: The Honorable Pamela G. Beidle, Chair,

I am writing as a private citizen to oppose SB117 (HB96) to add Krabbe Disease Leukodystrophy by statute to the Maryland Newborn Screening Panel.

I am the Dr. Frank V. Sutland Professor of Pediatric Genetics at Johns Hopkins University School of Medicine. I have served as the Clinical Director of Genetic Medicine since 2002. I also served on the Maryland State Advisory Council for Hereditary and Congenital Disorders (MACHCD) from 2000-2009, for the last 8 years as Chair.

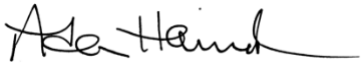
The composition of the MACHCD is mandated by statute since 1982 and includes representatives from the major academic institutions, as well as medical organizations, state legislators, and citizens who are caregivers for or affected by these conditions. This Council has an effective process in place to review and approve changes and additions to the Maryland Newborn Screening Panel. These issues are complex and require medical and public health expertise which is present on the Council and outside experts are also called in to present and discuss new conditions. This is not a process that should be subject to political pressure. There is no place for individual disease mandates through legislation.

The Council uses objective scoring criteria derived from the American College of Medical Genetics and Genomics newborn screening recommendations. MACHCD reviews conditions based on those reviewed and added to the federal Recommended Uniform Screening Panel (RUSP), which maintains a rigorous standard for review of data and evidence for the addition of new conditions. Per 2022 legislation, any condition added to the RUSP must be reviewed by MACHCD within a year, and if the Council does not recommend adding that condition to the Maryland Newborn Screening Panel, MACHCD must review the recommendation status annually. MACHCD reviews occur in open meetings and welcome presentations and discussion from all interested parties, who are given advance notice of meetings and the agenda.

Passage of this bill would set a dangerous precedent that would disrupt an effective and representative process for the consideration of new conditions to add to the Maryland Newborn Screening Panel. I strongly oppose this bill and any that would mandate inclusion of individual conditions by legislation.

Please don't hesitate to contact me with any questions.

Sincerely,



Ada Hamosh, MD, MPH

1 SB 117 -FIN- MDH- OPP.pdf
Uploaded by: Jason Caplan
Position: UNF

Maryland
DEPARTMENT OF HEALTH

Wes Moore, Governor · Aruna Miller, Lt. Governor · Laura Herrera Scott, M.D., M.P.H., Secretary

February 16, 2024

The Honorable Pamela Beidle
Chair, Senate Finance Committee
3 East, Senate Office Building
Annapolis, Maryland 21401

RE: Senate Bill 117 – Health - Newborn Screening Program - Krabbe Leukodystrophy – Letter of Opposition

Dear Chair Beidle and Committee Member:

The Maryland Department of Health (Department) respectfully submits this letter of opposition for Senate Bill (SB) 117 – Health - Newborn Screening Program - Krabbe Leukodystrophy. HB 96 mandates the addition of Krabbe leukodystrophy to the Department's Newborn Screening System.

Under current law, Health-General, §13–111 (d), the Department, in consultation with the State Advisory Council on Hereditary and Congenital Disorders (the Council), determines the screening tests that the Department's public health laboratory is required to perform. Deliberations within the Council involve extensive discussions on the inclusion of new conditions. Decisions are made using an objective scoring rubric, based on the American College of Medical Genetics Newborn Screening Guidelines, which carefully weighs the individual benefits of adding the condition versus cost to the state and any potential harm to families. In 2014 and 2018, during which time Krabbe Leukodystrophy was not on the HRSA Recommended Uniform Screening Panel (RUSP), the Council considered the addition of Krabbe Leukodystrophy and ultimately voted against its inclusion.[1,2]

Since the inception of MDH's Newborn Screening System in 1965, no condition has been added via legislation. Adding Krabbe leukodystrophy via legislation would circumvent the existing statutory process of requiring the State Advisory Council to deliberate and make recommendations on the detection and management of hereditary and congenital disorders.

The federal government has not yet added Krabbe Leukodystrophy to the Health Resources and Service Administration Recommended Uniform Screening Panel (RUSP). On January 30, 2024, the federal Advisory Committee on Heritable Disorders in Newborns and Children voted to recommend the addition of Infantile Krabbe Leukodystrophy to the RUSP. In order for the screening to actually be added to the RUSP, the Health and Human Services Secretary must choose to adopt the federal Advisory Committee recommendation, a step that has not yet occurred. The State Advisory Council on Hereditary and Congenital Disorders (Council) is committed to reviewing the addition Infantile Krabbe leukodystrophy to Department's Newborn Screen System as soon as the findings of the federal Advisory Committee are available.

[1] State Advisory Council on Hereditary and Congenital Disorders meeting minutes (4/1/14)

[2] State Advisory Council on Hereditary and Congenital Disorders meeting minutes (6/24/18)

The Department understands the need to review the latest data considered by the federal Advisory Committee and make a determination as soon as possible. For these reasons, the Department respectfully recommends that the committee vote unfavorably on SB 117.

If you would like to discuss this further, please do not hesitate to contact Sarah Case-Herron, Director of Governmental Affairs at sarah.case-herron@maryland.gov.

Sincerely,

Laura Herrera Scott, M.D., M.P.H.
Secretary

SB 117- ACHCD - LOS (Council position).pdf
Uploaded by: State of Maryland (MD)
Position: UNF

Maryland Department of Health
Advisory Council on Hereditary and Congenital Disorders

Wes Moore, Governor · Aruna Miller, Lt. Governor · Laura Herrera Scott, Secretary

January 30, 2024

Dear Chair Beidle

The Maryland Advisory Council on Hereditary and Congenital Disorders (MACHCD) opposes HB96 (SB117) to add Krabbe Disease Leukodystrophy by statute to the Maryland Newborn Screening Panel.

MACHCD has been in existence since 1982 and is mandated per statute to make recommendations to the Secretary of Health on which health conditions are appropriate for addition to the Maryland Newborn Screening Panel. Since the inception of Newborn Screening in Maryland, no disease has ever been added to the panel by direct legislative action. From the Council's founding statute, the MACHCD's diverse membership ensures that all viewpoints are represented in the recommendations of the council. MACHCD members include clinical experts on genetic, metabolic, and other conditions; general pediatrics; state legislators; and lay community members who are affected by congenital or hereditary disorders, either as affected individuals or caregiver/family members. The Council uses an objective scoring criteria derived from the American College of Medical Genetics newborn screening recommendations.

MACHCD reviews conditions based on the conditions reviewed and added to the federal Recommended Uniform Screening Panel (RUSP), which maintains a rigorous standard for review of data and evidence for additions of new conditions. Per 2022 legislation, any condition added to the RUSP must be reviewed by MACHCD within a year, and if the Council does not recommend adding that condition to the Maryland Newborn Screening Panel, MACHCD must review the recommendation status annually.

On January 30, 2024, the federal Health Resources Services Administration's Advisory Committee on Heritable Disorders in Newborns and Children voted to add early infantile Krabbe Disease to the RUSP. This vote is the first step to add Krabbe Disease to the RUSP as the Secretary of Health and Human Services must finalize the addition, similar to the process in Maryland.

MACHCD believes that conditions should not be added to the newborn screen by statute, bypassing the Council's process. The MACHCD process involves the review of complex medical and scientific information, including expert analysis of the disease-based literature and evidence of public health benefit. The Council spends significant time in consultation with disease experts, member discussion and questions for the experts and MDH laboratory and clinical follow-up staff, and scoring the condition against the objective scoring criteria.

As a body independent of the Maryland Department of Health, MACHCD feels no disease should be added to the panel via legislation. Instead, the state should continue to use the statutorily defined process that has been in effect for 42 years.

Sincerely,

Sincerely,

Jamie L. Fraser, M.D., Ph.D., DABMGG
Chair

Mailing Address: c/o Lauren Whiteman, 1770 Ashland Ave, Baltimore, Maryland 21205
Lauren.Whiteman@maryland.gov
443-681-3916

SB117_Krabbe_KennedyKrieger_LOI.pdf
Uploaded by: Emily Arneson
Position: INFO

Kennedy Krieger Institute

Bradley L. Schlaggar, MD, PhD
President and CEO
Zanvyl Krieger Faculty Endowed Chair

A comprehensive resource for children with disabilities

February 16, 2024

Senator Pamela Beidle
Chair, Senate Finance Committee
Maryland General Assembly
3 East Miller Senate Office Building
Annapolis, MD 21401

Re: Senate Bill 117 - Health - Newborn Screening Program - Krabbe Leukodystrophy

Dear Chair Beidle,

We respectfully submit this letter regarding SB117, proposed legislation that would add the rare neurogenetic disorder, Krabbe Leukodystrophy, to the newborn screening panel in Maryland.

As you know, this proposed legislation, would amend Maryland Statute 13-111 to specifically include Krabbe Leukodystrophy to the newborn screen. However, this statute already delineates a process that requires the Maryland Department of Health's Newborn Screening Program, in collaboration with the Advisory Council on Hereditary and Congenital Disorders, to determine whether to approve the inclusion of each condition listed in the U.S. Department of Health and Human Services' Recommended Uniform Screening Panel (RUSP).

We understand and appreciate the motivation for this proposed addition. On January 30, 2024, the Federal Newborn Screening Advisory Committee voted to add Infantile Krabbe Disease (IKD) to RUSP. Kennedy Krieger Institute agrees with and celebrates this determination by RUSP. **It is extremely important, in our opinion, that the State of Maryland is in alignment with newborn screening for all conditions listed under RUSP, including IKD. Further, it imperative that sufficient funding is allocated for the inclusion and implementation of IKD to the newborn screening process.** We also appreciate the extant language in 13-111, describing the required steps (and the required timeline for those steps) to be taken by the Maryland Department of Health, in consultation with the State's Advisory Council on Hereditary and Congenital Disorders, now that IKD has been added to RUSP.

For more than 40 years, Kennedy Krieger has been globally recognized as an authority on the study and care for patients diagnosed with a wide range of leukodystrophies. The Moser Center for Leukodystrophies at Kennedy Krieger provides comprehensive care to patients with leukodystrophies through an interdisciplinary approach, bringing together the fields of neurogenetics, genetic counseling, neurorehabilitation, endocrinology, and urology, along with physical, occupational, speech, and aquatic therapy. The Center works collaboratively with other leukodystrophy centers across the country and around the world.

Newborn Screening allows for the early detection of treatable rare genetic disorders, resulting in a dramatic improvement in the lives of young babies and children, as well as their families. Krabbe Disease is a type of neurodegenerative condition called a leukodystrophy. All leukodystrophies are rare inherited diseases that affect myelin (the "white matter" which insulates nerve cells in brain, spinal cord, and peripheral nerves). Recent data indicate that

707 North Broadway • Baltimore, Maryland 21205 • KennedyKrieger.org
main 443.923.9300 • **facsimile** 443.923.9317 • **tty** 443.923.2645 • **email** Schlaggar@KennedyKrieger.org

leukodystrophies affect approximately 1 in 7000 newborns. Krabbe Disease is caused by a genetic abnormality that results in in the buildup of a toxin called psychosine. Accumulation of psychosine leads to destruction of myelin. The most profound form of this disease is the infantile form, referred to as Infantile Krabbe Disease (IKD), which rapidly progresses leading to a vegetative state and death within the first 2 years of life. These newborns appear healthy and normal at birth but quickly develop abnormal eye movements, loss of motor skills, inability to feed, difficulty seeing and/or hearing, stiffness and spasms in the muscles, and seizures, leading to death. Currently the only available treatment to halt the rapid decline of IKD is hematopoietic stem cell transplantation. This treatment is only effective in changing disease trajectory if patients receive the stem cell transplant within the first few weeks of life before there is clear presence of neurological or developmental deficits. Therefore, it is imperative to identify children with IKD at birth.

Currently, eleven states include Krabbe Disease on their newborn screening panels. The eleven states are: New York, Missouri, Kentucky, Tennessee, Illinois, New Jersey, Ohio, Indiana, Pennsylvania, Georgia, and South Carolina. The cost of adding IKD to the newborn screening panel would be minor as many commercial screening kits already measure galactocerebrosidase (GALC) activity, the first-tier test for Krabbe Disease, in every sample and therefore at no additional cost. The additional cost for second tier psychosine testing in suspected cases would be relevant for only a miniscule fraction (i.e. 0.04%) of all screened newborns.

The purpose of newborn screening for the most dire diagnoses – such as IKD - is to be able to deliver rapidly hematopoietic stem cell transplantation within the first month of life. In addition to this standard treatment approach, there are multiple new gene therapy clinical trials in the pipeline to identify disease modifying treatments for the KD patient population. The option to undergo hematopoietic stem cell transplantation and the continued development of better gene-based therapies fail without newborn screening.

In sum, Kennedy Krieger celebrates the very recent addition of IKD to the federal Recommended Uniform Screening Panel. A fortunate consequence of this addition is that the extant language in Maryland statute 13-111 delineates the next steps in the process, and timeline for those next steps, for incorporating IKD into the Maryland newborn screen.

Sincerely,

Bradley L. Schlaggar, M.D., Ph.D.
President & CEO
Zanvyl Krieger Faculty Endowed Chair
Kennedy Krieger Institute

S. Ali Fatemi, MD MBA
Chief Medical Officer
Director, Moser Center for Leukodystrophies
Blum-Moser Endowed Chair
Kennedy Krieger Institute