February 5, 2024

The Honorable Joselyn Pena-Melnyk
Chair, House Health and Government Operations Committee
241 Lowe House Office Building
Annapolis, Maryland 21401

RE: HB096 – Health – Newborn Screening Program – Krabbe Leukodystrophy

Dear Chair Pena-Melnyk:

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