HB096 Health – Newborn Screening Program – Krabbe Leukodystrophy this information is:

The Honorable Joseline A. Pena-Melnyk

Chair, House Health, and Government Operations Committee

Room 241, House Office Building

Annapolis, MD 21401

In March of 2023, my rambunctious and vibrant red headed 19 month old stopped walking. By the end of the week if you tried to stand him up he would screech and scream at the top of his lungs. We got in for a physical therapy evaluation, they said low muscle tone that within a few weeks should improve. I left optimistic, but I wasn't prepared for the journey that would lie ahead of us. The following week, Arthur lost his ability to sit. I was shocked and scared. We went to the chiropractor to see if he thought there was a chance of a slipped disc in his lower back. After a thorough exam, he said he felt it was more neurological. I immediately called our pediatrician. Originally they said they couldn't see Arthur for 1 week. I expressed how it simply couldn't wait, and miraculously at the same time someone called and canceled their appointment for 1030 in the morning with our primary doctor. When she came in, she noticed he a significant decline from 2 months prior. She started to say she would send us to ortho and as she thought about it she said and maybe neurology. Immediately I knew we would be waiting a long time as pediatric neurology generally has a 6-12 month waiting list. But as she started to leave the room she turned back to check his reflexes and her face dropped (she didn't know it at the time that she didn't check them properly, but if she had he wouldn't be here) she felt his reflexes had declined significantly (when in reality he had hyper reflexes). We were sent to the children's hospital expecting a tumor on spinal chord or cancer (which would have almost been preferable to our actual diagnosis). After all the tests and procedures all with no conclusive answers we were sent home pending genetic testing results. We were told they suspected a leukodystrophy or mitochondrial disease. We continued on to our daughter's 11th birthday trip to Universal studios for the following weekend thinking it might be our last trip as a family.

On our first day there we got a call from the doctor, Krabbe Disease. Most of the conversation was a blur, but we heard stem cell transplant and prognosis of 1 year post symptoms. The rest of the week was spent researching and trying to find treatment. We came home early from out trip to begin planning praying we could receive treatment that could buy him more time here with us. After fighting for a week with insurance we were finally approved to be seen and evaluated by the NDRD team for rare diseases in Pittsburgh.

By the time we arrived here Arthur could no longer sit independently, his toes were chronically pointed and hands were balled into fists. Dr. Rajan then told us the reality, she didn't feel like he was a good candidate for treatment. Mentally he was thriving, but his body was declining fast. She predicted by August he would have lost his sight and possibly his ability to breathe, all ability to move arms and legs, and swallow on his own. She then said based on the progression she imagined he would pass before the year was up if we chose not to proceed with treatment. She felt it would be best to bring him home and enjoy him until he was called home to be with Jesus. Transplant would be rough and cause his body to decline faster and progress the disease faster until he engrafted which would be his baseline. He would continue to decline for a few months before improving back to baseline over the next year. Our hearts were shattered. She would give us 24 hours to decide if we still wanted to meet with the transplant team. We prayed and felt we would always question ourselves if we didn't. We met the transplant team, we were told he wouldn't move arms or legs, wouldn't be able to swallow or possibly breathe on his own, BUT he felt we could preserve his smile and a nice belly laugh. After testing and MRI's we drove home only to be called 2 hours after departure that insurance approved everything and we needed to be back Monday to be admitted. Wednesday we had a G-tube and picc lines placed. Friday we began 2 weeks of chemotherapy and were transplanted June 2nd.

While recovery was rough finally on June 23rd we were considered engrafted. We were inpatient for 4.5 months and outpatient for 1.5 months. 10 hours from home and daughters.

The week before Thanksgiving we were finally released to come home.

I am writing this today to express how vital it is to have early diagnosis. Most child pass away simply because diagnosis came too late. If my son's medical team hadn't been diligent he, too would now most likely be dead. I urge you to implement Krabbe on your newborn screening. All children deserve a quality of life, and parents deserve the choice to transplant or not. Arthur was late infantile and we got to see him talk, walk, and develop a beautiful personality before the disease took it all from him. Parents shouldn't be robbed of that choice.