PEDIATRIC BLOOD & MARROW TRANSPLANT PROGRAM
STEM CELL LABORATORY
ROBERTSON CLINICAL & TRANSLATIONAL CELL THERAPY PROGRAM
CAROLINAS CORD BLOOD BANK

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To Whom It May Concern:

I am writing to provide written testimony in support funding of the Pennsylvania's Newborn Screening Program implementation of Act 148 of 2014. I write as an expert in the care and treatment of pediatric patients with Krabbe Disease.

Since 1990 I have served as the director of the Pediatric Blood and Marrow Transplant (PBMT) Program at Duke University Medical Center. The PBMT program has transplanted over 60 infants and children with early infantile, late infantile and juvenile Krabbe Disease. The program has also transplanted and followed approximately 21 infants with early infantile Krabbe disease transplanted with unrelated donor umbilical cord blood transplantation in the first 2 months of life. Over 80% of these children survive more than 10 years and all survivors have improved qualities of life post-transplant. All children are cognitively normal, but most have some motor impairments as they get older. Children transplanted in the first 30 days of age have superior outcomes compared to those transplanted over 30 days of age. Ten years or more post-transplant, approximately 1/3 of children ambulate normally or with assisted devices while 2/3 are wheelchair bound. While transplant is not a cure, it definitely improves outcomes and provides opportunity for additional interventions over time.

Babies with Krabbe Disease experience tremendous suffering in infancy and early childhood, for as long as they survive. They develop feeding difficulties in the first few months of life, followed by loss of milestones (smiling, head support, eye contact), develop extreme irritability (day and night), blindness, spasticity, seizures, respiratory compromise and death. Parents seek medical consultation to learn what is wrong with their baby, but usually go from doctor to doctor for months on end before receiving a diagnosis. One of the hardest things is that the babies don't sleep, the family doesn't sleep and mothers can't comfort their babies. When the diagnosis is made in a baby with symptoms, it is too late for intervention with transplantation. Also, in some cases, the family has subsequent pregnancies with affected babies. These could have been avoided if the family was aware of their risk to conceive a baby with early infantile Krabbe disease (EIKD).

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Hematopoietic stem cell transplantation after high dose chemotherapy is the only treatment available for patients with Krabbe Disease at this time. Experimental treatments giving additional cells into the spinal fluid, gene therapy and enzyme therapy are under development and may improve prognosis in the future.

I have treated over 60 babies and children with Krabbe disease. As mentioned above, infants with EIKD treated under 30 days of age have the best outcomes. Toddlers with late infantile disease do very well if transplanted before symptoms develop. Children with juvenile disease also do well even after presenting with mild symptoms. There are no placebo controlled trials because the disease is too rare and some consider it unethical to let children die since outcomes without transplant are well described.

In conclusion, I strongly support the funding of Pennsylvania's Newborn Screening Program implementation of Act 148 of 2014. This will enable screening for Krabbe disease and enable treatment of presymptomatic patients, informed subsequent family planning in affected families, and avoid the anguish families experience searching for a diagnosis only to learn that it is too late to help their baby.

Please do not hesitate to contact me with any questions at kurtz001@mc.duke.edu or 919-668-1119.

Sincerely,

Joanne Kurtzberg, MD
Jerome Harris Distinguished Professor of Pediatrics & Pathology
Chief Scientific Officer and Medical Director, Robertson Clinical & Translational Cell Therapy Program
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Director, Carolinas Cord Blood Bank at Duke