Kennedy Krieger Institute

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A comprehensive resource for children with disabilities

February 7, 2024

Delegate Joseline A. Pena-Melnyk
Chair, Health and Government Operations
Maryland General Assembly
Room 241 House Office Building
Annapolis, MD 21401

Re: House Bill 96 - Health - Newborn Screening Program - Krabbe Leukodystrophy

Dear Chair Pena-Melnyk,

We respectfully submit this letter regarding HB96, 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

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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,

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