



State of Wisconsin
Department of Health Services

Scott Walker, Governor
Linda Seemeyer, Secretary

August 30, 2016

Thu-Thoa Trinh, MD
1000 North Oak Street
Marshfield, WI 54449

Dear Dr. Trinh,

Thank you for submitting a nomination on April 9, 2015, to add Krabbe disease to the newborn screening panel of conditions. We appreciate and share your concern for children and families who are profoundly affected by this terrible disease.

When we receive nominations to add a condition to the Wisconsin panel, we follow a careful process to ensure they are thoroughly reviewed by doctors and scientific experts in Wisconsin, using established criteria. You can read more about this process, and the committees involved, at <https://www.dhs.wisconsin.gov/newbornscreening/process-additions.htm>.

In November, 2015, a subcommittee of Wisconsin physicians who specialize in caring for children with metabolic disorders such as Krabbe reviewed your nomination and the available evidence. These doctors, who recommended against adding Krabbe to the newborn screening panel at this time, noted the following:

"Krabbe disease is a progressive neurological disorder. By the time children are symptomatic there is no effective therapy. For those of us who have watched the decline of children afflicted by this disorder it is heart wrenching. For parents the feeling of powerlessness and the suggestion that an earlier diagnosis may have improved the outcome lead to a desire to prevent other parents from suffering in this way. Consequently it is logical to wish to identify this disorder by newborn screening and offer the hope of potential therapy.

"However, as identified in the literature and evidence review attached, there remains significant problems with unacceptably high false positive rate of proposed screening methods. This problem is compounded by difficulty in distinguishing true positive cases from false positives with subsequent molecular or enzymatic testing. Furthermore since the evidence review there has been no further published studies demonstrating that HCST [hematopoietic stem cell transplantation] provides an improvement in quantity or quality of life over supportive care. Therefore at this time the metabolic subcommittee cannot support the addition of Krabbe disease to the state of Wisconsin newborn screening panel."

In December, 2015, a larger committee of Wisconsin newborn screening specialists reviewed the nomination and also recommended against adding Krabbe to the newborn screening panel at this time.

In January, 2016, the Secretary's Advisory Committee on Newborn Screening (SACNBS) reviewed the nomination, as well as the recommendations of the earlier committees. Dr. Joanne Kurtzberg, professor of pediatrics and pathology at Duke University, called in to describe the work she is doing in treating

children with Krabbe. In addition, the SACNBS heard moving public testimony, including testimony from families whose children have been affected by Krabbe.

On August 2, 2016, The SACNBS recommended that the Krabbe Disease nomination proposal be tabled pending the acquisition and development of more information, and encouraged a resubmission at a time when the unanswered questions could better be addressed.

One of the criteria that must be met for a condition to be added to the Wisconsin panel is that there must be a safe and effective treatment to prevent serious health consequences. As of January, there were no published studies showing effective treatment for Krabbe. Dr. Kurtzberg described promising treatments, and we hope that in the very near future her results will be reviewed by her peers and formally published.

Another criterion is that there must be a good plan for how to manage cases of children who have positive screening results, but may not have the disorder. As the Wisconsin subcommittee identified, in many cases it is currently not possible, even after extensive testing, to determine whether a positive Krabbe screen means a child will later develop problems. This leaves parents and their children in a state of anxious uncertainty, with no way to find answers.

Based on the recommendation of the SACNBS, the Department has made the difficult decision that Krabbe should not be added to the Wisconsin newborn screening panel at this time. We encourage you or others to resubmit the nomination when additional published evidence becomes available for effective treatment and approaches for managing false positive screens. We look forward to a time when children born with Krabbe can lead full and healthy lives.

Thank you for your interest and support for the Wisconsin Newborn Screening Program.

The full report of the SACNBS is enclosed, and is also posted on the DHS website's *Krabbe Disease – Nomination Process* page (<https://www.dhs.wisconsin.gov/newbornscreening/krabbe-nomination-process.htm>).

Sincerely,

A handwritten signature in black ink, appearing to read "Linda Seemeyer", with a long horizontal flourish extending to the right.

Linda Seemeyer
Secretary

Enclosure