DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
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February 17, 2010

Ms. Jacque Waggoner
Chief Executive Officer
Hunter's Hope Foundation
PO Box 643
Orchard Park, NY 14127

Dear Ms. Waggoner:

The Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) appreciates your nomination of Krabbe Disease for inclusion in the Committee's recommended uniform newborn screening panel for State newborn screening programs. Acknowledging the importance of the condition and the status of the test development and treatment, the Committee requested its external evidence review workgroup to formally review the evidence from both published and unpublished data regarding screening, diagnosis, and follow-up care for Krabbe. The attached report from the comprehensive evidence review was presented to the Committee and thoroughly discussed at its September 2009 meeting.

The Committee found Early Infantile Krabbe Disease (EIKD), the "the better defined end of the spectrum, would benefit from early diagnosis and intervention" thus indicating a benefit from screening. However, Committee members also found that "substantial harm is possible (either from testing and/or identification; from treatment/other interventions, or both)". The Committee determined that there are some specific gaps in the evidence that must be addressed in order to further consider adding Krabbe disease to the recommended uniform newborn screening panel. Therefore, the Committee voted to recommend not adding the condition. The wording of that motion is as follows: "*There is insufficient evidence for the Committee to make a recommendation to add the condition to the core panel, and there is insufficient evidence of potential net benefit to lead the Committee to want to make a strong recommendation regarding pilot studies*."

The Committee identified the following evidence gaps:

(1) **EIKD, The Condition:** need consensus about the case definition of what constitutes Early Infantile Krabbe Disease (EIKD)

(2) **Test for EIKD, Screening and Diagnosis:** there is a need for additional information about the testing algorithm for EIKD. It is important to ascertain whether testing for Krabbe disease would be a stand alone test or done with multiplex testing, in part because of the cost implications.

(3) **Treatment for EIKD:** More information is needed about the specific benefits of Hematopoietic Stem Cell Transplant (HSCT) to treat patients and what mutations would benefit most from HSCT.

The Committee understands that additional states will be adding Krabbe Disease to their NBS panel. It is anticipated that some data will be available from those states that could address the evidence gaps, and highly recommended that the data be made available to the Committee and its external evidence review workgroup. A potential strategy to addressing the gaps would be to develop a research partnership with the National Institutes of Health's newly established Newborn Screening Translational Research Network, under the leadership of the American College of Medical Genetics.

The Committee will reconsider its recommendation after the new evidence addressing the above issues are made available for the Committee's review and re-evaluation.

Sincerely yours,

/s/

R. Rodney Howell, M.D.
Chairman

Enclosure- Final Krabbe External Evidence Review Workgroup Report

cc: Joanne Kurtzberg, M.D.
Michele Caggana, Sc.D., FACMG
Maria Luisa Escolar, M.D.
Ms. Micki Gartzke