



## DEPARTMENT OF HEALTH AND HUMAN SERVICES

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Secretary's Advisory Committee on Heritable  
Disorders in Newborns and Children  
5600 Fishers Lane, Room 18A19  
Rockville, Maryland 20857  
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<http://www.hrsa.gov/heritabledisorderscommittee/>

Elliott Vichinsky, M.D  
Children's Hospital – Oakland  
Hematology Department  
747 52nd Street  
Oakland, CA 94609

Dear Dr. Vichinsky:

The Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) appreciates your nomination of Hemoglobin H 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the Committee requested its external evidence review workgroup to formally review the evidence from both published and unpublished data regarding screening, diagnosis, and follow-up care for Hemoglobin H disease. The enclosed report from the comprehensive evidence review was presented to the Committee and thoroughly discussed at its May 2010 meeting.

The Committee found regarding Hemoglobin H type of alpha-thalassemia that “the issues with hemoglobin H are similar to other diseases the Committee has considered previously. Hemoglobin H can be a late onset disease, which may present clinically between 0-73 years of age.” They recognized that diagnosis is easier in the neonatal time frame and that “people running screening programs, regardless of the recommendations of the Committee, face an ethical decision about deciding to ignore evidence that is right in front of them every time they perform IEF.” However, Committee members felt that they had “not heard any compelling evidence to suggest that hemoglobin H belongs on the screening panel. There was no compelling clinical need presented.” Examples of clinical utility would include how many patients receive a splenectomy, at what age they receive them and whether or not they require transfusions afterwards. The Committee determined that there are some specific gaps in the evidence that must be addressed in order to further consider adding Hemoglobin H disease to the recommended uniform

newborn screening panel. Therefore, the Committee voted not to add Hemoglobin H to the panel and to place it as Category 4 “Additional Evidence Needed.”

- Evidence gaps identified the following:
  - What proportion of children would benefit from condition-specific treatment? There is a lack of follow-up data on screen positive children.
  - What is the variation in prevalence across the United States?
  - Does early identification improve the health of identified children?
  - What is the threshold for moving a target from secondary status to one of the core targets?
  - In terms of infrastructure, what are the expectations for newborn screening laboratories, public health clinicians, and families if there is a move from secondary to a primary target?

The Committee understands that California, Hawaii and other states will continue to routinely screen for and identify Hemoglobin H disease. 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.  
Chairperson

Enclosure:

Final Hemoglobin H External Evidence Review Workgroup Report