DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
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September 1, 2009

The Honorable Kathleen Sebelius
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Sebelius:

The Advisory Committee on Heritable Disorders in Newborns and Children (the Committee) is charged with advising the Secretary of the Department of Health and Human Services in areas relevant to heritable conditions in newborns and children such as newborn and child screening, counseling, or health care services for newborns and children having or at risk for heritable disorders. This letter is to inform the Secretary of the Committee's progress to date in their evidence review deliberations and in other Committee actions relevant to newborn and child screening programs.

Legislation under the Newborn Screening Saves Lives Act 2008 indicates the Committee:

1. Provide recommendations, advice, or information on certain diagnostic and screening activities;

2. Provide such recommendations, advice or information as may be necessary to enhance, expand or improve the ability of the Secretary to reduce the mortality or morbidity in newborns and children from heritable disorders;

3. Make systematic evidence-based and peer-reviewed recommendations that include the heritable disorders that have the potential to significantly impact public health for which all newborns should be screened, including secondary conditions that may be identified as a result of the laboratory methods used for screening;

4. Develop a model decision-matrix for newborn screening expansion, including an evaluation of the potential public health impact of such expansion and periodically update the recommended uniform screening panel, as appropriate, based on such decision-matrix;

I have enclosed a Committee report that outlines the Committee's evidence review process (Tab A). At its last meeting on September 24-25, 2009, the Committee received an evidence report on Krabbe Disease from the Evidence Review Workgroup and made the following recommendation to the Secretary:

- **Recommend not adding the condition (Krabbe Disease) to the core panel now**. *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.*

I will send a letter to the nominators of Krabbe Disease about the Committee's decision to recommend not adding Krabbe Disease to the Committee's recommended newborn screening panel at the present time. The letter will indicate the areas in which the evidence pertaining to Krabbe Disease had been found to be deficient, especially regarding the condition, the test (screening and diagnosis), and the treatment. The Committee will recommend that the nominators of this condition address these issues before returning the nomination.

As part of its evidence review process, the Committee considered the nomination of Hemoglobin H disease (alpha thalassemia) for an evidence review. As a group, thalassemias are the most common single gene disorders of humans, and thalassemia protects against malaria and is most prevalent where malaria is endemic. Thalassemias are due to impaired production of globin chains, creating an uneven balance between hemoglobin's α (alpha chains) and β (beta chains). There are several types of α-thalassemia with varying degrees of clinical significance. Hb H disease, which is caused by the deletion or inactivation of three α globin genes and an excess γ-chain (or Hemoglobin Bart's) or β chain (depending on the age at which an individual is tested), are the most significant clinically.

The Committee accepted the recommendation of its Nomination Review and Prioritization Workgroup that Hemoglobin H disease receive a formal evidence review by the External Evidence Review Workgroup. The Committee requested that the evidence review consider that the condition would move from the secondary panel to the core newborn screening panel and address specific questions such as the value of early detection and the value of treatment, during the first year of an infant's life.

The Committee also considered a report (Tab B) from its Subcommittee on Education and Training to improve genomic education of primary care physicians

involved in maternal and child health, and unanimously adopted two recommendations, that the Secretary, HHS:

- Develop and fund a "Learning Collaborative" in genetics and primary care training to support increased genetic literacy amongst primary care providers.

- Provide additional resources to increase public awareness of the newborn screening system.

There are two other policy areas under consideration by the Committee. A draft paper entitled "Considerations and Recommendations for a National Policy Regarding the Retention and Use of Dried Blood Spot Specimens after Newborn Screening" includes state practices for the use and storage of residual biological samples remaining after the newborn is screened and recommendations to the Secretary. "Newborn Screening and Health Care Reform: Report of U.S. Secretary of Human Services' Advisory Committee on Heritable Disorders in Newborns and Children", provides information on how health care reform might improve the quality and efficiency of public health newborn screening programs and equal access to those services for the benefit of the families that they serve. Reports addressing these two arenas are currently under Committee review.

In addition, I announced that two new Committee workgroups are being formed. One workgroup will address information and material from emerging data sets and registries, and newborn screening codes related for heritable disorders with participation from the Health Resources and Services Administration and other Federal agencies such as the Centers for Disease Control and Prevention and the National Institutes of Health. The second workgroup will prepare a draft response to the report of President Bush's Council on Bioethics (now disbanded) for Committee review and publication.

In conclusion, please know that the Committee stands ready to be of service to you to help strengthen newborn screening programs. Effective newborn screening programs can play an important role in improving the health of our children.

Sincerely yours,

R. Rodney Howell, M.D.
Chairperson

Enclosures:
Tab A: Committee's Evidence Review Process
Tab B: Committee's Education and Training Subcommittee Report