



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable
Disorders in Newborns and Children
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March 30, 2009

The Honorable Charles E. Johnson
Acting Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Johnson:

Title XXVI of the Children's Health Act of 2000, "Screening for Heritable Disorders," established a program to improve the ability of States to provide newborn and child screening for heritable disorders by adding sections 1109, 1110, and 1111 to the Public Health Service (PHS) Act. The Secretary of Health and Human Services (HHS) was directed under section 1111 of the PHS Act to establish an Advisory Committee on Heritable Disorders in Newborns and Children (Committee). On April 24, 2008, the "Newborn Screening Saves Lives Act of 2008" reauthorized the PHS Act and amended the programs and activities under sections 1109 and 1111, and added Sections 1112, 1113, 1114, 1115 and 1116.

The Committee provides technical information to the Secretary for the development of policies and priorities that will enhance the ability of State and local health agencies to provide newborn and child screening, counseling, or health care services for newborns and children having or at risk for heritable disorders. Legislation under the Newborn Screening Saves Lives Act 2008 indicates the Committee shall, amongst other duties:

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;

This letter is to inform the Secretary of the Committee's progress to date in their evidence review deliberations.

Over the past year, the Committee has developed its nomination and evidence review process. Thus far, six conditions have been sent to the Committee for consideration of an evidence review for addition to the Committee's recommended uniform screening panel. The six conditions include Krabbe Disease, Severe Combined Immunodeficiency (SCID), Pompe Disease, Fabry Disease, Niemann-Pick disease, and Spinal Muscular Atrophy (SMA).

- Niemann Pick Disease, Fabry Disease and SMA were deemed by the Committee as not ready for review by the external evidence review workgroup.
- On October 1, 2008, the Committee voted unanimously to recommend not adding Pompe Disease to the Committee's recommended uniform screening panel, but encouraged additional specific studies. The Committee sent a letter to the nominator of Pompe Disease outlining the additional specific studies to be conducted before the nomination of Pompe Disease will be reconsidered by the Committee.
- On February 26, 2009, the Committee voted to recommend not adding SCID to the Committee's recommended uniform screening panel but encouraged additional specific studies. The Committee sent a letter to the nominator of SCID outlining the additional specific studies to be conducted before the nomination of SCID will be reconsidered by the Committee.
- The preliminary report on the nomination of Krabbe disease to the Committee's recommended uniform screening panel will be presented by the external review workgroup during the May 12, 2009 Committee conference call.

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