

Advisory Committee on Heritable
Disorders in Newborns and Children
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October 2008

The Honorable Michael O. Leavitt
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Leavitt:

The Advisory Committee on Heritable Disorders in Newborns and Children (Committee), is charged with advising the Secretary of the U.S. Department of Health and Human Services in areas relevant to heritable conditions in children, especially newborn screening. At its October 12, 2008 meeting, the Committee reviewed and discussed the American Health Information Community's (AHIC) report and use case for Newborn Screening. The Committee voted unanimously to endorse the efforts of the Subgroup on Newborn Screening of the AHIC Personalized Health Care Workgroup to develop a newborn screening use case and resource guide for the use case.

Newborn screening is a public health measure to detect and prevent or reduce the consequences of serious, often life-threatening, insidious, and rare genetic and congenital disorders for over 4 million infants in the US. Within the newborn screening system, the chain of information flow involves a complex interaction among public and private laboratories, hospitals and primary care offices, subspecialty referral centers, state registries, and others. The development of the Newborn Screening Use Case is an important step in the process of determining the information technology requirements and the opportunities for using communication interoperability to improve the timeliness and completeness of the newborn screening process.

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 newborn screening panel. The six conditions include Severe Combined Immunodeficiency (SCID); Pompe; Krabbe; Niemann-Pick; Fabry and Spinal Muscular Atrophy (SMA). Niemann Pick and Fabry Diseases were deemed by the Committee not to be ready for an evidence review; the Committee decisions are pending for SCID and SMA. At its October meeting, the Committee voted unanimously to recommend **not adding** Pompe Disease to the Committee's uniform newborn screening panel and recommended additional studies to be conducted before the nomination of Pompe Disease would be reconsidered by the Committee.

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,

/s/

R. Rodney Howell, M.D.
Chairperson
ACHDNC