DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
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February 25, 2010

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 including newborn and child screening, counseling, and health care services for newborns and children having or at risk for heritable disorders.

The Health Resources and Services Administration's (HRSA) Maternal and Child Health Bureau (MCHB) commissioned the American College of Medical Genetics (ACMG) in 2001 to convene an expert panel to outline a process of standardization of outcomes and guidelines for state newborn screening programs, including a recommended uniform panel of conditions to include in state newborn screening programs. The ACMG expert panel was asked to conduct an analysis of the scientific literature on the effectiveness of newborn screening and gather expert opinion to delineate the best evidence for screening specified conditions and develop recommendations focused on newborn screening, including but not limited to the development of a uniform condition panel. It was expected that the analytical endeavor and subsequent recommendations be based on the best scientific evidence and analysis of that evidence. Upon review of the final ACMG report to HRSA/MCHB and the public comments to that report, in 2005 the Committee endorsed the report and its recommendations.

Since this time the Committee has developed its nomination and evidence review process. Thus far, nine conditions have been nominated to the Committee for consideration of an evidence review for addition to the Committee's Recommended Uniform Screening Panel: Krabbe Disease, Severe Combined Immunodeficiency (SCID), Pompe Disease, Fabry Disease, Niemann-Pick Disease, Spinal Muscular Atrophy, Hemoglobin H Disease, Hyperbilirubinemia/Kernicterus, and Critical Cyanotic Congenital Heart Disease. Three nominated conditions were initially deemed by the Committee as not ready for evidence review (Fabry Disease, Niemann-Pick Disease, and

Spinal Muscular Atrophy) and three nominated conditions have had a complete evidence review but were not approved for addition to the Recommended Uniform Screening Panel (Krabbe Disease, Severe Combined Immunodeficiency (SCID), and Pompe Disease). The remaining three other nominated conditions: Hemoglobin H Disease, Hyperbilirubinemia/Kernicterus, and Critical Cyanotic Congenital Heart Disease, are currently moving through the evidence review process. At the Committee's January 2010 meeting, the nominators of SCID requested a re-evaluation by the Committee of SCID and related T-cell lymphocyte deficiencies based on the availability of additional evidence.

When developing its recommendations to the Secretary, the Committee considers the nature of the science itself underlying the potential additions of the technology and the heritable conditions to the Recommended Uniform Screening Panel and public health implications of implementation. It is with these issues in mind that the Committee recommends a tiered approach to the implementation of screening for SCID and related T-cell lymphocyte deficiencies. In addition, because SCID and related T-cell lymphocyte deficiencies are rare in the States, and in order to gain the knowledge necessary through an iterative implemental development of infrastructure needed for ongoing research, evaluation, surveillance, education, and training for screening for SCID and related T-cell lymphocyte deficiencies, the Committee therefore recommends to the Secretary:

- The addition of SCID to the uniform panel, and related T-cell lymphocyte deficiencies to the list of secondary targets as a comprehensive entity, with the understanding that the following activities will also take place in a timely manner.
    - The National Institutes of Health shall fund surveillance activities to determine health outcomes of affected newborns with any T-cell lymphocyte deficiency receiving treatment as a result of prospective newborn screening;
    - The Health Resources and Services Administration shall fund the development of appropriate education and training materials for families and public health and health care professionals relevant to the screening and treatment of SCID and related T-cell lymphocyte deficiencies.
    - The Centers for Disease Control and Prevention shall develop and distribute to performing laboratories suitable dried blood spot specimens for quality control and quality assurance purposes.

This is the first condition determined to be ready for addition to the Committee's Recommended Uniform Screening Panel since 2005. It is a milestone for this Committee and represents the success of the Committee's evidence review system. Thank you for your consideration of this important topic.

Sincerely yours,

R. Rodney Howell, M.D.
Chairperson