DEPARTMENT OF HEALTH AND HUMAN SERVICES

Discretionary Advisory Committee on
Heritable Disorders in Newborns and Children
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June 3, 2013

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

Dear Secretary Sebelius:

The Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (the Committee) is charged with making systematic evidence-based and peer-reviewed recommendations that include heritable disorders that have the potential to significantly impact public health for which all newborns should be screened. During the Committee's May 2013 meeting, the Committee reviewed the objective evidence report for the nominated heritable disorder –Pompe disease (also known as glycogen storage disease type II or acid maltase deficiency). Based on this report which included a public health impact assessment and Committee deliberations, the Committee voted to recommend that you update and expand the Recommended Uniform Screening Panel (RUSP) to include the addition of Pompe Disease.

Pompe disease is an autosomal recessive disorder with a prevalence rate of approximately 1 in 28,000. Pompe ranges from an infantile form that can result in significant morbidity and death in early childhood to a late-onset form with a variable presentation that causes progressive weakness and respiratory failure.

The Committee feels strongly that there are significant benefits in screening for Pompe disease. Data shows that screening for Pompe, as opposed to clinical identification alone, results in earlier diagnosis and treatment of the infantile form of the disease. Enzyme replacement therapy has been shown to significantly modify the course of the infantile form of Pompe disease and earlier treatments with enzyme replacement therapy result in better outcomes for affected infants. The screening tests have a high sensitivity and specificity in detecting infants with Pompe disease. The addition of Pompe disease to the RUSP will also allow for more research to occur to examine the impact of early treatment for late onset cases thus helping to minimize the prolonged and often painful search for a diagnosis faced by adults with late onset of Pompe disease.

Nonetheless, the Committee recognizes that although it is feasible for states to implement Pompe disease screening, the majority of newborn screening programs will need a period of time to obtain the necessary equipment and staff as well as establish treatment referral

networks in order for successful implementation to occur. In addition, I, as Chair of the Committee, hope that currently funded newborn screening research projects, such as the NIH/NICHD Newborn Screening Translational Research Network, can be involved with States which are capable of being the first to include Pompe disease screening and assist them in developing evidence based approaches for the standardization of testing protocols and the evaluation and management strategies for persons found to have late onset Pompe disease.

The Committee believes with high certainty that screening for Pompe disease will lead to significant net benefits for infants born with this rare condition.

Sincerely yours,

Joseph A. Bocchini, Jr., M.D.
Chairperson