



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Advisory Committee on Heritable Disorders in
Newborns and Children
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www.hrsa.gov/advisory-committees/heritable-disorders

July 25, 2019

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

Dear Secretary Azar:

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) provides advice, technical information and recommendations regarding genetic disorders, newborn screening, and childhood screening. The objective of the Advisory Committee is to enhance states' abilities to reduce morbidity and mortality in newborns and children who have, or who are at risk for, genetic disorders. ACHDNC is charged with making systematic evidence-based and peer-reviewed recommendations regarding conditions for inclusion on the Recommended Uniform Screening Panel (RUSP). The RUSP is a list of conditions recommended by the Secretary of the Department of Health and Human Services for states to screen for at birth as part of their state universal newborn screening (NBS) programs. In addition, the ACHDNC is responsible for advising the Secretary on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and standards. This charge includes providing the Secretary guidance and information on health care services for newborns and children who have or are at risk for heritable and other life-threatening disorders. Under this charge, ACHDNC developed a report on *Medical Foods for Inborn Errors of Metabolism: The Critical Need to Improve Patient Access* (attached).

President Trump in his Address to a Joint Session of Congress on February 28, 2017 highlighted the importance of providing breakthrough therapies for rare congenital disorders. He introduced Megan Crowley, a 20-year-old sophomore at Notre Dame, who was diagnosed with Pompe Disease when she was 15 months old. The infantile form of Pompe Disease was a uniformly fatal genetic condition. The President pointed out the successful efforts of her father to develop the drug that saved her life.

The ACHDNC reviewed available evidence at its May 2013 meeting and found that Pompe Disease met the evidence-based criteria necessary to recommend addition to the RUSP. The review included information on reliable screening tests, which could be incorporated into a newborn screening laboratory, the development of a successful enzyme replacement therapy and the clinical impact of early treatment on the outcome of affected individuals. The ACHDNC therefore recommended to Secretary Sebelius that Pompe Disease be added to the RUSP. On March 2, 2015, Secretary Burwell accepted the recommendation. At present, eighteen states/territories screen for Pompe and another twelve are pursuing adding Pompe to their newborn screening panel.

While Pompe Disease is an excellent example of how NBS can dramatically alter the prognosis for individuals with an included condition, access to treatment for several conditions on the RUSP is problematic. Specifically, persons with inborn errors of metabolism, which are not treated by drug therapy, but by “medical foods” to avoid permanent morbidity and mortality, often experience difficulties in obtaining insurance coverage for the required therapy throughout their lifetime.

The Food and Drug Administration (FDA) defines medical foods per section 5(b)(3) of the Orphan Drug Act (21 U.S.C. 360ee(b)(3)), to be “a food which is formulated to be consumed or administered enterally under the supervision of a physician and which is intended for the specific dietary management of a disease or condition for which distinctive nutritional requirements, based on recognized scientific principles, are established by medical evaluation.”

The ACHDNC has been concerned for a number of years that the medical foods required to maintain the health of persons with inborn errors of metabolism are not covered by medical insurance, as are the drugs, which are necessary for treatment for the other conditions on the RUSP. The ACHDNC completed a review of the medical food reimbursement in the United States and prepared a white paper on the subject, which highlights the urgent need to address the discrepancies in coverage that exist from state to state. Many families have inadequate resources to provide needed medical foods for affected children, and young adults often struggle to maintain lifesaving treatment when coverage is not available.

Phenylketonuria (PKU), as the first screened condition, serves as a paradigm disorder to illustrate the remarkable inconsistency in access to medical food across the United States. Without access to prescribed medical food, individuals with PKU suffer the consequences of toxic elevations in phenylalanine and its metabolites in the blood and brain. These abnormal levels have intellectual, developmental, behavioral, and mental health consequences that include difficulties in school, work, and relationships. Moreover, some affected individuals experience long-term neurologic damage related to failure to maintain dietary control. For individuals with other inborn errors of metabolism, lack of access to medical foods leads to failure to grow and possibly a metabolic crisis with permanent neurologic damage or death.

We hope this report provides your office with the information needed to address this unsolved problem for our most vulnerable populations of children. We look forward to working with you in your efforts to improve the lives of newborns and children, and their families.

Sincerely yours,

/s/

Cynthia M. Powell, MD, MS, FACMG, FAAP
Chairperson

/s/

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

Enclosure:

Report – *Medical Foods for Inborn Errors of Metabolism: The Critical Need to Improve Patient Access*

cc: Catharine Riley, PhD, MPH
Designated Federal Official
Health Resources and Services Administration