



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Discretionary Advisory Committee on
Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 18A19
Rockville, Maryland 20857
www.hrsa.gov/heritabledisorderscommittee

December 16, 2014

The Honorable Sylvia Mathews Burwell
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Burwell:

It is an honor to welcome you to your new position as Secretary of Health and Human Services. In April 2012, the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) was established under the Public Health Service Act (PHS), 42 U.S.C. 217a: Advisory councils or committees. The DACHDNC fulfills the functions previously undertaken by the former Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) established under the Section 1111 of the Public Health Service (PHS) Act, 42 U.S.C. 300b-10, as amended in the Newborn Screening Saves Lives Act of 2008 (Act). The mission of the DACHDNC is to reduce morbidity and mortality in newborns and children who have, or are at risk for, heritable disorders. The committee advises the Secretary of the U.S. Department of Health and Human Services on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and standards.

During the September 11-12, 2014 meeting, the DACHDNC reviewed and discussed a report, *Succinylacetone as Primary Marker to Detect Tyrosinemia Type I in Newborns and its Measurement by Newborn Screening Programs*, which provides data on using succinylacetone as a primary marker to detect Tyrosinemia type 1 (TYR 1) as well as information on the different TYR 1 screening practices currently used in newborn screening laboratories across the nation.

The DACHDNC's Laboratory Standards and Procedures Subcommittee first identified the inconsistent implementation of screening for TYR 1. TYR 1 is a genetic disorder and is caused by an autosomal recessive fumarylacetoacetate hydrolase deficiency. It is the most severe form of Tyrosinemia with symptoms appearing within the first few months of life. Symptoms include failure to thrive, diarrhea, vomiting, yellowing of the skin and whites of the eyes (jaundice), cabbage-like odor, and increased tendency to bleed (particularly nosebleeds). If left untreated, most infants die of liver failure in the first year of life. Research shows that intervention is most effective if treated within the first month of life.

The DACHDNC determined that the advice and information detailed in the enclosed report have a high potential impact and ability to improve practices and outcomes for individual states. The Committee voted to officially support the report and issue the following formal recommendation for Secretarial consideration:

The Secretary of Health and Human Services should facilitate a national dialogue among federal and state stakeholders on the benefits of measuring succinylacetone in dried blood spots to improve the specificity of newborn screening for Tyrosinemia type I, a condition on the Recommended Uniform Screening Panel.

We hope that the Centers for Disease Control and Prevention will play a major role in facilitating this national dialogue and recommend that various stakeholders be a part of the dialogue including representatives from laboratories and state newborn screening programs. On behalf of the DACHDNC, we look forward to receiving your decision on this DACHDNC recommendation. Thank you for your consideration on this important topic. Please know that the DACHDNC stands ready to be of service to you to help strengthen the newborn screening programs that play such an important role in improving the health of the Nation's children.

Sincerely yours,

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

Enclosure:

Report - Succinylacetone as Primary Marker to Detect Tyrosinemia Type I in Newborns and its Measurement by Newborn Screening Programs

cc: Debi Sarkar, M.P.H.
Designated Federal Official
Health Resources and Services Administration