Advisory Committee on Heritable Disorders in Newborns and Children

Advisory Committee on Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 18W68
Rockville, Maryland 20857
301-443-2521– Phone
www.hrsa.gov/advisory-committees/heritable-disorders

December 7, 2020

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

Dear Secretary Azar:

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC or Committee) provides advice, technical information and recommendations regarding genetic disorders and newborn and childhood screening. The objective of the ACHDNC is to enhance states' abilities to reduce morbidity and mortality in newborns and children who have, or who are at risk for, genetic disorders. The Committee also makes systematic evidence-based and peer-reviewed recommendations regarding conditions for inclusion on the Recommended Uniform Screening Panel (RUSP): a list of conditions recommended by you, 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 programs.

During the February 8, 2018 meeting, the Committee reviewed the evidence-based report for the nominated heritable disorder – spinal muscular atrophy (SMA). Based on this report and deliberations on all associated clinical data, testing platforms, available treatments, benefits and harms, an assessment of the public health systems impact, and public comment, the Committee voted to recommend to you the following: Expand the RUSP to include the addition of SMA due to homozygous deletion of exon 7 in Survival Motor Neuron 1.

On July 2, 2018, the ACHDNC received notification that you accepted the Committee's recommendation to include SMA on the RUSP. In your letter, you asked the Committee, to provide a report, "…describing the status of implementing newborn screening for SMA and clinical outcomes of early treatment, including any potential harms, for infants diagnosed with SMA."

In collaboration with subject matter experts, the Committee developed a report detailing states' experiences with implementation of screening for SMA and its impact on newborns with the condition. On December 1, 2020, the Committee voted to approve the *Review of Newborn Screening Implementation for Spinal Muscular Atrophy Final Report.* On behalf of the Committee, I am pleased to provide you with the report and am available to answer any questions you may have.

Sincerely yours,

/s/

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