United States Senate
WASHINGTON, DC 20510

April 26, 2018

Secretary Alex Azar
Department of Health and Human Services
200 Independence Avenue, SW
Washington, D.C. 20201

Dear Secretary Azar,

We write regarding the recent recommendation by the Health Resources and Services Administration (HRSA) federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) to add spinal muscular atrophy (SMA) to the Recommended Uniform Screening Panel (RUSP) for newborn screening. We urge you to swiftly review the recommendation.

Adopted in 2010, the RUSP is a list of conditions the Department of Health and Human Services (HHS) recommends states include in their newborn screening programs. The ACHDNC advises the HHS Secretary on the most appropriate application of universal newborn screening tests, including which conditions to add to the RUSP. Once a disease is added to the RUSP, many states work quickly to update their screening panels to include the additions.

The importance of the RUSP in encouraging newborn screening cannot be overstated. Each year, approximately four million newborns in the United States are screened for many serious and debilitating conditions that are not clearly apparent at birth. Newborn screening ensures that these disorders are identified early, allowing for prompt initiation of treatments and helping achieve the best possible health outcomes for the affected child. In many cases, a delayed diagnosis until symptoms present can be disastrous, and potentially fatal.

On February 8, 2018, the ACHDNC recommended adding screening for SMA to the RUSP; after receiving the recommendation, you have up to 120 days to accept or reject it. SMA is a leading genetic cause of infant death. SMA affects the nerve cells in the spinal cord that control motor function, robbing many patients of the ability to walk, eat, or breathe. It can affect any race or gender. The most severe type of SMA, Type 1, is also the most common type. Based on symptom onset alone, babies with SMA typically receive a diagnosis within their first six months

of life and rarely survive beyond two years without intensive and invasive therapies, including permanent mechanical ventilation.

In December 2016, the Food and Drug Administration (FDA) approved the first drug treatment for SMA. Clinical study data for this treatment appears to support initiation of treatment in the pre-symptomatic phase of the disease. Screening newborns for SMA will shorten diagnosis time and provide greater likelihood that treatment will start before symptom onset, thereby improving the prospect that a child will not experience serious developmental delays or premature death.

Due to the critical nature of this disease and the benefits of early diagnosis, we urge you to act expeditiously in reviewing ACHDNC's recommendation.

Thank you for your consideration of this matter.

Sincerely,

Orrin G. Hatch
United States Senator

Edward J. Markey
United States Senator