January 16, 2025

The Honorable Richard C. (Rip) Sullivan Jr.
Virginia House of Delegates
General Assembly Building
201 North 9th Street, Room 908
Richmond, Virginia 23219

Re: HB 1782 – Newborn Screening Requirements

Dear Delegate Sullivan,

As patient advocacy organizations representing individuals diagnosed with rare diseases and their families in Virginia, we write to express our support for HB 1782, regarding newborn screening. This bill, if passed, would ensure the state at a minimum considers national recommendations when determining which conditions newborns are screened for at birth.

Each of the rare diseases we represent impacts fewer than 200,000 Americans. For many of these patients, only a few physicians nationwide have the specialized knowledge needed to provide them with a diagnosis and treatment. Too often, limited access to specialty care contributes to a delayed diagnosis and irreversible disease progression. On average, it takes more than 6 years and about 17 medical interventions for an individual with a rare disease to receive an accurate diagnosis.[1] When screened at birth for pediatric-onset conditions that are known to be devastating, yet treatable when acted upon, newborns receive opportunities for lifesaving diagnoses and medical interventions.

In anticipation of the advent of scientific innovations that yield new medical treatments and interventions, rare disease communities work for decades to develop infrastructure to support newborn screening and early intervention programs. Such infrastructure includes diagnostic testing, clinical care guidelines, provider and patient education resources, and a follow up support system. These resources and data are then reviewed as a part of the evidence gathered by the evidence review panel of the federal advisory committee to consider whether the benefit of screening for a condition weigh against the potential harms in the context of public health. This list is the Recommended Uniform Screening Panel (RUSP).

Rare disease patient communities that have worked to develop national infrastructure for families once identified through newborn screening rely on states to proactively review and adhere to these federal advisory committee recommendations. Yet we recognize that each individual state must consider condition addition within the context of the resources available within a particular state. Thus, the state RUSP Alignment legislative approach; to ensure that conditions that have met the rigorous evidentiary requirements of the federal RUSP are implemented within the tailored timelines and resource requirements of each state.

Virginia is a leader in the nation screening for 35 of the 38 RUSP conditions. HB 1782 aims to codify existing practices in Virginia's newborn screening program that have proven to be effective. The legislation also creates a streamlined process for the state health officials and the legislature to work together to review new RUSP conditions, ensuring that babies do not go undiagnosed due to unnecessary

[1] The National Economic Burden of Rare Disease Study, EveryLife Foundation for Rare Diseases, everylifefoundation.org/burden-landing/

administrative delays. We ask you to support HB 1782 and look forward to working with you to ensure that Virginia's newborns have access to life-changing and potentially life-saving diagnoses.

Sincerely,

The EveryLife Foundation for Rare Diseases