February 14, 2025

The Honorable Louise A. Lucas
Chair, Senate Committee on Finance and Appropriations
201 N 9th Street, Senate Room A
Richmond, VA 23219

Dear Chair Lucas and members of the Committee on Finance and Appropriations,

On behalf of the one-in-ten individuals Americans living with one of the over 10,000 known rare diseases, the National Organization for Rare Disorders (NORD) thanks you for the opportunity to provide comments on House Bill 1782 (HB 1782).

NORD is a unique federation of voluntary health organizations dedicated to helping people with rare diseases and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services. We believe that all individuals with a rare disease should have access to high quality, affordable health care that is best suited to meet their medical needs.

HB 1782, if passed, would direct the Virginia Department of Health to evaluate conditions that appear on the Recommended Uniform Screening Panel (RUSP) and begin rulemaking to add conditions to the Commonwealth's newborn screening panel within 12 months of their addition to the RUSP. It would also require the Department of Health to determine annually whether disorders not included on the Commonwealth's newborn screening panel should be reevaluated for inclusion. Finally, the Department would be required to submit an annual report to the General Assembly detailing the disorders included, evaluated, not recommended for inclusion, and not recommended for reevaluation. NORD urges you to support HB 1782 and advance the bill favorably out of the Committee on Finance and Appropriations.

Newborn screening is a vital public health program with a long history of successful operation in the United States, improving the health of many children through timely diagnosis and treatment. Screening began in the 1960s when Dr. Robert Guthrie developed a blood test for a condition called phenylketonuria (PKU). PKU is a serious metabolic disorder that causes brain damage if it is not detected and treated very early in life. Children with PKU appear healthy at birth, but they are born without an enzyme necessary to break down certain proteins. As a result, an amino acid called phenylalanine builds up in the body causing permanent damage. Before Dr. Guthrie's blood test, children with PKU weren't diagnosed until after they had suffered irreversible brain damage. The blood test allowed health care providers to detect PKU shortly after birth, enabling earlier treatment and avoiding serious health complications caused by the condition. Now, nearly 4 million newborns across the country are screened each year for a number of rare conditions that, like PKU, can cause permanent disability or death in the absence of early detection and treatment. Babies born with these serious conditions typically appear healthy at birth, so screening is critical to ensure newborns who need it receive early care, treatment, or

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intervention as early as possible. Over 14,000 babies are found to have a serious rare disorder through newborn screening each year. In many cases, these early diagnoses are lifesaving and as science progresses, an increasing number of conditions are being added to the screening panel, helping to save additional lives.

Newborn screening programs are operated at the state level, and the specific conditions on each state's newborn screening panel varies across the country. To guide state newborn screening programs, the Health Resources and Services Administration's (HRSA) Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) maintains a list of conditions recommended for screening called the Recommended Uniform Screening Panel. To be added to the RUSP, a condition must meet all of the following criteria: 1) be a serious medical condition; 2) have a clear and well-described case definition; 3) have a screening process that has high analytical validity; 4) have a screening process that is clinically useful; 5) have well-defined treatment protocols; and 6) have data available demonstrating that population-based screening works well to find newborns with the given condition.[1] Virginia currently screens for 35 of the 38 conditions on the RUSP, and does not have a defined timeline for when conditions added to the RUSP must be added to the Commonwealth's newborn screening panel.

By not screening newborns for all conditions on the RUSP, the Commonwealth risks missing critical information about newborn Virginians' health, and children with conditions that are not on the newborn screening panel can suffer preventable, irreversible health complications as a result NORD supports the timely addition of RUSP conditions to state newborn screening panels and the process laid out in HB 1782. We encourage the Committee to support this legislation and advance it favorably to the full Senate.

Once again, on behalf of the Virginia rare disease community, we thank you for the opportunity to provide comments on HB 1782. For any questions, please feel free to contact Allison Herrity at aherrity@rarediseases.org.

Sincerely,

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CC: Leah Barber, Director of Grassroots Advocacy, NORD