EVERYLIFE FOUNDATION FOR RARE DISEASES
COMMUNITY CONGRESS

Dear Chairwoman Powell and members of the Advisory Committee for Heritable Disorders in Newborns and Children,

On behalf of the over 30 million Americans living with rare diseases, and as co-chair of the EveryLife Foundation's Newborn Screening and Diagnostics Working Group, I am pleased to offer the following comments to inform the Advisory Committee's ongoing conversations about the review process for new RUSP nomination packages. The EveryLife Foundation for Rare Diseases is a nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments, and cures.

The Community Congress is a forum for collaboration across stakeholders, representing over 200 individual rare disease patient advocacy organizations in addition to over 90 other healthcare and biotechnology organizations. Our Newborn Screening and Diagnostics Working Group is dedicated to ensuring that the rare disease community receives the earliest possible access to lifesaving diagnostic opportunities through newborn screening and other diagnostic tools.

We understand that the periodic evaluation of the RUSP nomination process is necessary to ensure that standards are current and rigorous. We appreciate that the Advisory Committee sought out input from the patient community multiple times during this review process and are continuing that practice today. Our communities work for many years with relevant partners and experts to develop a newborn screening system that includes the dried blood spot screening, confirmatory testing, educational materials, and follow-up infrastructure for our respective disease communities. This requires investment in developing care standards and screening tools, conducting pilots, and then leading the compilation of a nomination package that meets the evidentiary requirements for the RUSP.

To inform your activities we offer the following comments.

On the Condition Nomination Form, what additional information would better inform the Committee?

Recognizing the significant workload of the Advisory Committee, and the pipeline of conditions that may be nominated to the Committee in the near term, we urge you to consider the following recommendations for additional information on the Condition Nomination Form:

- The assessment of the benefit of screening for new conditions should accept a degree of uncertainty regarding the amount of data available following the approval of a treatment or the availability of an intervention, and include all sources of information, such patient community insights. Parallels can be drawn from the review and regulation of treatments, where FDA weighs such factors in order to speed the availability of new treatments for serious or life-threatening diseases to address unmet medical needs.
- The use of long-term data plays a vital role in understanding the potential impact of conditions being considered for RUSP nomination. The use of long-term newborn

screening data can help to close health equity gaps, improve health outcomes, inform earlier clinical care guidelines, and provide important data to guide health policy. The creation of a central database for review of long-term data would provide the Advisory Committee the ability to track incoming data for conditions planning to submit a RUSP package. Many patient organizations are leading longitudinal data collection efforts within adjacent ecosystems and would be critical and eager partners in this endeavor.

What information is difficult to obtain?

If a child's disease is not picked up via newborn screening, they often go years without an accurate diagnosis. Opportunities to study a treatment or intervention in infants and young children are limited as a result. In the absence of early detection, it is challenging to obtain data for certain decision-making criteria requested to demonstrate the benefit of earlier diagnosis. That same data is often required when submitting a RUSP Nomination Form:

- Successful RUSP nominations require prospective, population-based pilots that may cost millions of dollars and take several years to complete. It may not be feasible for many patient organizations to shoulder the financial responsibility of building a framework for a newborn screening pilot. Other diseases are so rare that conducting a state pilot that satisfies existing decision-making criteria may not be feasible.
- The same challenges associated with developing a treatment for a rare disease will exist when assessing the benefit of newborn screening. Disease rarity, heterogeneity, and other disease-specific considerations may impact the ability to assess the benefit of newborn screening within a population.

The decision matrix is a tool to assist the Committee in making decisions. Are there suggestions for additions or edits on the decision matrix?

When approving a condition, the Committee must consider that not all rare diseases will follow the same trajectory. Some diseases, when left untreated, may result in death within the first years of life. Many other rare diseases are progressive and equally devastating, with irreversible decline beginning early in life. In such cases, clinical outcomes may take years to measure and newborn screening provides a gateway to improve current treatments and develop new ones that will stop that decline. We urge the Committee to update the decision matrix to account for the variability in disease trajectory when considering the benefits to newborns.

We appreciate that the COVID-19 pandemic has placed even greater demands on the precious commodities of time and resources on our newborn screening leaders, and we are especially grateful for your unwavering dedication to our rare disease patient communities. The EveryLife Foundation and the membership of our Community Congress Newborn Screening and Diagnostics Working Group stand ready to support your work and we look forward to engaging with you over the coming months.

Sincerely,

The EveryLife Foundation for Rare Diseases