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.

Newborn screening serves as the first step in reaching a diagnosis, and preventing the many heartbreaking, costly, and physically devastating years, known as the diagnostic odyssey that is associated with many rare diseases. Our communities work for many years with relevant partners and experts to develop a newborn screening system and infrastructure around our disease community, invest in developing care standards and screening tools, and conducting pilots, and then work to help lead the compilation of a nomination package that meets the evidentiary requirements for the RUSP. We understand that the periodic review of this process is necessary to ensure that standards are current and rigorous. As you proceed, we hope the review and implementation of an updated evidence review process continues to be conducted in a manner that is transparent and inclusive of patient community members as key experts. We'd like to thank you for today's public comment period dedicated to this topic.
To inform your activities we offer the following comments.

- As the Advisory Committee seeks to update your review process for newly nominated conditions, while also considering a new process for assessing implementation and outcomes for conditions previously added to the RUSP, it is important to ensure that any new functions of the Advisory Committee can occur without sacrificing the pace or quality of the review of newly nominated conditions. Both of these Committee roles are important to the future of newborn screening, but neither should inhibit the ability for the other to occur.
- As an organization dedicated to supporting patient advocacy groups that are leading newborn screening programs, we are aware that numerous strong nominations will be submitted to the Advisory Committee in the coming years. As you prepare for an increasing number of RUSP nomination packages, we urge the Advisory Committee to develop a strategy to ensure that no condition's review is delayed due to the lack of resources to review new packages.
- As you are aware, communities work for many years to develop newborn screening systems and develop the evidence required for a RUSP nomination. There are currently multiple patient advocacy organizations that are developing their RUSP nomination

package using the evidentiary requirements and evidence review process. We request that the Advisory Committee considers these communities and current research projects when considering any changes made to the evidence review process. In addition, we ask that you consider a transition period, rather than a specific transition date, for enabling a phasing of any evidentiary requirement changes and updates.

We are pleased that the Advisory Committee is requesting feedback about the educational materials that will accompany the update to the review process. These materials will help ensure the newborn screening community understands and adapts to the changes and that no community is left behind because they were unaware of the updates to the evidence review process.

- The revisions to the review evidence review process will impact stakeholders across the newborn screening system. Any change to the data required for addition to the RUSP will impact how newborn screening researchers, including academic, private, and state public health laboratories, design their studies in preparation for a RUSP package. Any potential review of current RUSP conditions will require additional oversight and data reporting for state newborn screening programs. For these reasons, we suggest the Advisory Committee to create a variety of educational materials for newborn screening stakeholders, identifying changes to the evidence review process and how those changes impact specific communities throughout the entire newborn screening system.
- Creating a collection of education materials will help to ease the transition from the current evidence review process to the updated review process. To accomplish these educational goals, we encourage the Advisory Committee to establish a multistakeholder working group – to include representatives from the patient community – to inform the development and dissemination of these materials.

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