Dear Chairman Calonge 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 opportunities to enhance engagement with the stakeholder community. My name is Dr. Kim Stephens and I am pleased to be here today as the President of Project Alive, the Co-Chair of the EveryLife Foundation Community Congress Newborn Screening and Diagnostics Working Group, and as a parent advocate.

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 100 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.

Through our newborn screening efforts, the EveryLife Foundation works with many of the diverse stakeholders that comprise the newborn screening ecosystem every day. We appreciate Chair Calonge's acknowledgement of the vital role we stakeholders play during the August meeting – and we know that it is our patient advocates, clinical providers, researchers, public health officials, and policymakers that make the newborn screening system one of the most successful public health programs in the country.

To ensure the Committee remains representative of the newborn screening ecosystem's diverse stakeholders, we encourage the Committee to provide more formalized opportunities for engagement. To this end, we have the following suggestions for consideration of the Committee:

- We recommend that as the composition of the Committee is considered, professional diversity among Committee members and representation of the ecosystem be reflected.
- We request the Committee once again add a patient advocate as a committee member, so that they may properly represent the patient experience on the Committee.
- We ask the Committee take time to respond to public testimony in order to develop a deeper dialogue between the Committee and the various stakeholders that make up the newborn screening system.
- We request that the Committee add a public comment section to their quarterly meetings dedicated to advocates speaking about conditions currently within the RUSP nomination review process. This practice, which the Committee already utilizes when a condition is up for final vote for RUSP nomination, will ensure the public has an increased amount of time to communicate their perspectives to the Committee.
- Finally, we ask that the Committee include patient advocates in panel presentations to ensure presentations and subsequent conversations that occur at the Committee meetings are properly represented by the patient experience. As you are aware, most newborn screening programs

are driven with patient community involvement. That fact is not reflected in Committee agendas and proceedings.

We appreciate the immense responsibility the Committee takes on in recommending which life-altering conditions newborns should be screened for and understand that the Committee is limited in the number of conditions you can review. We ask that as the Committee continues forward, you actively communicate your concerns regarding resources and other limitations within newborn screening; but not allow those concerns to dominate conversations. In turn, as a patient advocacy community, we will continue to work with policymakers to provide more resources and reduce workforce barriers, so that all newborns may realize the lifesaving benefits of newborn screening.

Thank you for the opportunity to speak with the Committee today, and for your unwavering dedication to our rare disease patient community. The EveryLife Foundation and members of our Community Congress Newborn Screening & Diagnostics Working Group look forward to our continued engagement over the coming months.

Sincerely,

The EveryLife Foundation for Rare Diseases