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, the EveryLife Foundation for Rare Diseases is pleased to offer the following comments to inform the Committee's ongoing conversations about the evidence review process for RUSP nomination packages. The EveryLife Foundation 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 one of four working groups of the Community Congress which is dedicated to ensuring that the rare disease community receive the earliest possible access to lifesaving diagnostic opportunities through newborn screening and other diagnostic tools.

We applaud the Committee's recommended addition of MPS-II to the Recommended Uniform Screening Panel (RUSP). Once accepted and implemented, the addition of MPS-II will provide the approximately 38 MPS II babies born annually in the U.S. the opportunity to access timely life-saving early diagnosis and treatment. However, we wish to note that during the Committee's review of the MPS-II nomination, there were discussions of topics we feel were outside the scope of the MPS-II review. During the EveryLife Community Congress Newborn Screening Working Group meeting that followed the ACHDNC February meeting, members expressed these concerns directly. Comments centered on worries that some of the discussion by the Committee strayed from the task of examining the quality of evidence of the MPS-II nomination and its impact on the public health system, and instead focused on challenges presented by the current healthcare system. While such discussions are important and we are grateful for the Committee's commitment to addressing these challenges, we enjoin the Committee to ensure such important discussions do not become barriers to enabling new, worthy conditions from being added to the RUSP.

As a rare disease community, we appreciate that the Committee is working to prepare for an anticipated increase in RUSP nominations. As the Committee navigates this increased demand, we remind the Committee of the importance of formally including the patient community voice in the review process and the importance of expanding review capacity.

- As the Committee begins the selection and onboarding process of new members, we request the Committee commit to seeing all 15 member positions filled. By fully staffing the Committee, discussions of nominations, their review, and the broader newborn screening ecosystem will benefit from the greater expertise and personal experience that a fully constituted Committee will provide. As the Committee prepares to onboard these new members, we ask for increased

transparency of the onboarding process. As an organization dedicated to supporting patient advocates, we want to ensure that the patient voice is represented throughout this onboarding process.

- We also request the Committee include two clinical representatives from the technical evidence review committee in the final review discussion to help answer questions as they come up during the Committee's final deliberation. The inclusion of these experts will allow for key insights into the impact newborn screening would have on a disease community and would allow them to respond to any specific inquiries about the condition that may arise.
- As the Committee considers how to handle the increasing number of RUSP nominations, we strongly encourage the Committee to focus on expanding their capacity to review these conditions as opposed to focusing on how to prioritize nominated conditions. We worry that focusing on prioritization of conditions could limit the RUSP and close it off to other worthy rare diseases.
- The Committee has stated that they are capable of supporting only two evidence reviews per year. We understand that while the Committee may have limited ability at the moment to increase that number, we also ask that they provide increased transparency concerning the docket of pending nominations. We recommend that transparency include a brief synopsis at each meeting of all pending nominations with respect to dates and where they are in the process, to include the dates and other relevant information regarding submission to HRSA, assignment to the Nomination Prioritization Working Group, those undergoing ERG-review, and those scheduled for ERG-review discussion and vote. Patient organizations prepare for many years building evidence and developing their nomination package, and require a clear timetable of when their RUSP review could potentially take place after submission.
- We are thankful that over the last few ACHDNC meetings, the Committee has highlighted challenges associated with newborn screening outside the RUSP. Presentations discussing various workforce issues have helped to highlight how many professionals are connected to newborn screening. As more treatments for rare diseases are developed, the newborn screening community will continue to look at ways to address these current challenges. However, we request that these conversations occur outside of an individual RUSP nomination review process and continue to be a separate activity for the ACHDNC.

We appreciate the Committee's dedication to meeting the increasing demands on the nation's newborn screening program, 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