

**MDA Comments to Secretary's Advisory Committee (Aug 25, 2016)**  
**Speaking Time: 3 minutes**

Thank you for the opportunity to address the Committee. My name is Kristin Stephenson, and I serve as vice president of policy and advocacy for the Muscular Dystrophy Association, and I am here today representing the tens of thousands of families and individuals who are living with muscular dystrophy, Spinal Muscular Atrophy (SMA), and other neuromuscular disorders.

MDA is a national non-profit organization dedicated to saving and improving the lives of people living with neuromuscular disease. To this end, MDA funds research; supports more than 150 care centers nationwide; and champions policies and programs important to those we serve—such as the robust and critical public health program that is newborn screening.

We are pleased that Pompe has been added to the recommended panel, and aim to work together with the community to see other neuromuscular diseases included as well, such as SMA and Duchenne muscular dystrophy (DMD).

With considerable advancements in the therapeutic pipeline and with current studies in process to develop the requisite data to support an application for nomination to the RUSP, we believe both SMA and DMD will prove strong candidates for addition to the panel—and we urge the committee to support those nominations as they are submitted.

Therapeutics to treat the underlying cause of both disorders are moving forward and a well established nationwide network of care centers exists to provide follow up care to infants as they are identified through the screening process. While there is much work to be done, MDA is committed to this readiness. We are also proud to be part of robust and significant collaborative efforts to move newborn screening forward for both SMA and DMD, that includes researchers, clinicians, advocacy groups, industry and other key stakeholders. As a community, we are preparing for newborn screening for these disorders.

We have recently entered an exciting phase as researchers have identified the genetic causes of many neuromuscular diseases and precision medicines are in development to target the underlying cause of disease.

SMA is the leading genetic cause of death for infants. The pace of therapy development in SMA is unprecedented, as the causative gene was only discovered a decade ago, and we are now seeing the first human trials testing therapies that target the underlying cause of disease. There are currently seven therapies in clinical trials for SMA, with over a dozen other approaches nearing the clinic.

Innovative strategies such as gene therapy and antisense oligonucleotide therapy are also being tested in SMA patients and are showing encouraging data. Recently, a large Phase 3 trial was halted, due to the trial meeting its primary endpoint in an interim analysis. We hope in the coming months to witness the first filing for a new drug application for SMA, a pivotal step in the fight to get an approved, effective treatment.

Similarly, for DMD, There are now 30 drugs in clinical development for DMD, and the FDA is currently reviewing a drug that would modify the underlying disease for approximately 13% of those living with Duchenne.

Time is of the essence in implementing newborn screening for SMA, DMD, and other neuromuscular diseases where early treatment is the best—and perhaps only—impactful approach to alter the natural progression of the disorder.

The significant drug development efforts ongoing for SMA and DMD are encouraging and we hope many of the other disorders covered under MDA's umbrella will follow in a similar path. In addition to SMA and Duchenne, there are infantile forms of other types of muscular dystrophy and neuromuscular disorders that could benefit from early intervention, and we look forward to sharing more information with the committee on these and other disorders in the future.

Thank you for your time today and for your dedication to reducing morbidity and mortality in newborns and children who have, or are at risk for, heritable disorders.