Oral Comment to the
Advisory Committee on Heritable Disorders in Newborns and Children
August 2020

On behalf of Parent Project Muscular Dystrophy (PPMD), I would like to thank the Committee for providing me with the opportunity to address you today. My name is Annie Kennedy and I serve as the Chief of Policy & Advocacy for the EveryLife Foundation for Rare Diseases -- and I am pleased to be presenting here today as a representative of the National Duchenne Newborn Screening Pilot Steering Committee.

For the last 6 years, Parent Project Muscular Dystrophy (PPMD) has been leading a national effort to build a Newborn Screening (NBS) infrastructure for Duchenne in the U.S. aimed at developing the evidence to support Duchenne newborn screening.

Building on the work led by Dr. Jerry Mendell in the Ohio state NBS pilot which concluded in 2012, PPMD endeavored to learn from the best practices of that pilot and refine the systems further such that they could be replicated in a state with a high birthrate – and eventually nationwide. In Dr. Mendell’s study, nearly 60,000 babies were screened throughout the state and 6 children with Duchenne were positively identified, establishing evidence for a 2-tier screen at birth for Duchenne within the US newborn screening system.

Our Duchenne effort has convened experts and established the partnerships required to research, pilot, and implement nationwide NBS for Duchenne. PPMD's Duchenne NBS efforts have included the expertise and input of experts and leaders within NIH, HRSA, FDA, CDC, the American College of Medical Genetics and Genomics (ACMG), the broader newborn screening community, and the Duchenne community.

Our efforts also included a study to determine which approach to CK screening had appropriate analytical/clinical validity and utility for use by a public health laboratory. These efforts were conducted in collaboration with the California Department of Health, PerkinElmer, UC Davis, UCLA, Stanford, and UCSF. We are delighted that the yield from these efforts have now been FDA approved and applied to current newborn screening pilot efforts.

We have also collaborated with the CDC and the American Academy of Pediatrics (AAP) to develop diagnostic tools and resources for primary care providers and families.
As a result of all of these collaborative efforts --

**In September of 2019, we initiated a Duchenne Newborn Screening Pilot in New York State.**

The Duchenne Newborn Screening pilot program was designed to set up, validate and conduct a consented pilot screen for infants born at select hospitals in New York State and utilizes tools, resources, and expertise at PPMD, the [Newborn Screening Translational Research Network (NBSTRN)](https://www.nbsrn.org), and the New York State Department of Health.

Our pilot is being funded through a unique model in which PPMD has convened a pre-competitive consortium of biopharmaceutical industry partners with a commitment to early diagnosis and intervention in Duchenne. Consortium members currently include PPMD, PTC Therapeutics, Sarepta Therapeutics, PerkinElmer, Solid Biosciences, and Pfizer, Inc. In addition, the pilot is being guided by a Steering Committee comprised of representatives from federal agencies, provider groups, and representatives from key Duchenne stakeholder communities.

While early pilot data will be shared by Dr. Norma Tavakoli at the APHL Newborn Screening Symposium later this fall, our first 9 months of the pilot – conducted in ground zero of the global pandemic – have demonstrated the unwavering commitment of our partners. It also has demonstrated that the infrastructure we worked so long to build has well served our partners.

We are so grateful to the leadership within New York State – within the state laboratories, the birthing centers and primary care provider sites. But most importantly, we are grateful to all those who are working with us to ensure that babies identified through this program are receiving the most immediate, expert, and comprehensive follow up care possible.

To prepare for this moment, PPMD has been working for nearly 2 decades on efforts aimed at readying the landscape for newborn screening efforts in Duchenne. These efforts have included:

- leadership on the Newborn Screening Saves Lives Reauthorization Act,
• annual Duchenne-specific language within Appropriations and Report Language to ensure that our federal partners are focused on Duchenne NBS efforts,

• and leading the National Duchenne Newborn Screening Initiative which has included the development of published care standards for newborns, ethical considerations for Duchenne NBS, and the publication of a Roadmap to Newborn Screening for Duchenne.

**Today**, we are exceptionally grateful to the families, experts, and partners who have helped us to get this far – and who have agreed to ‘lean in’ even further as we move the resources we’ve developed into this New York State pilot.

With three recently approved therapies – and a research pipeline filled with potential therapeutic interventions – newborn screening will provide optimal opportunities for care and treatment in Duchenne.

Our Duchenne Newborn Screening Pilot in New York State is an exciting and critical next step in improving outcomes for children with Duchenne.

Thank you.