Nomination package submitted to add Duchenne to the Recommended Uniform Screening Panel (RUSP)

PPMD'S PRESS RELEASE

Parent Project Muscular Dystrophy (PPMD), a nonprofit organization leading the fight to end Duchenne muscular dystrophy (Duchenne), announced that the organization submitted a nomination package to add Duchenne to the Recommended Uniform Screening Panel (RUSP) to the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). The RUSP is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their newborn screening (NBS) programs. Having a disorder be added to the RUSP shows that there is clear evidence of benefit to the babies and their families for newborn screening of that condition.

PPMD has been building the infrastructure for early identification of newborns with Duchenne—including a robust therapeutic pipeline, regulatory infrastructure, and clinical care network and guidelines—since its inception and preparing for the implementation of NBS for Duchenne for almost a decade. A mandatory element of this process was the development and support of a large pilot, which was completed in October of 2021. That pilot screened more than 36,000 babies born in New York State over two years. Four babies were confirmed to have Duchenne/Becker muscular dystrophy, and one baby was identified as a carrier female.

The New York State pilot was funded and led by a consortium of Duchenne industry partners and PPMD with a commitment to early diagnosis and intervention in Duchenne. Consortium members include PerkinElmer, Pfizer, Inc., PTC Therapeutics, Sarepta Therapeutics, Solid Biosciences, Wave Life Sciences, and PPMD. The pilot was guided by a Steering Committee comprised of representatives from healthcare professional groups, expert clinicians, and Duchenne stakeholder communities including the American Academy of Pediatrics, the Centers for Disease Control and Prevention, the EveryLife Foundation for Rare Diseases, the Genetic Alliance's Expecting Health, the Newborn Screening Translational Research Network (NBSTRN), and New York State.

The RUSP package submission will now initiate the ACHDNC's review process. In this review, the ACHDNC will analyze the evidence presented about the benefit of NBS for Duchenne. Considerations will include the availability of an effective test to identify newborns, the availability and need for treating young children with Duchenne, and the ability of state laboratories to do the screening. The review process for RUSP consideration typically takes more than a year and requires two key votes of experts in NBS to move forward.

Niki Armstrong, PPMD's Newborn Screening Program Manager explains that the submission of the RUSP nomination package is a great achievement, and another exciting leap towards the organization's goal of having Duchenne added to NBS programs across the country:

“While every state chooses which diseases are included in that state’s newborn screening panel, many states use the RUSP to guide their decisions. Most states also have their own separate process for evaluating which disorder to add, and some states may choose to screen for diseases even if they are not on the RUSP. Advocacy at the state level will be important to PPMD’s ongoing NBS efforts to promote implementation within each state.”

“EARLY DIAGNOSIS WILL MEAN EARLY INTERVENTION”

– PPMD's Pat Furlong

The RUSP nomination has been led by PPMD, with the Muscular Dystrophy Association (MDA) as a co-sponsor. Recognizing the need for early diagnosis, many other partners within the community including other patient advocacy and industry organizations have provided letters of support. The RTI Early Check DMD pilot, a pilot in North Carolina led by the nonprofit research institute RTI International with funding from the MDA and Sarepta Therapeutics, and the Brigham Women’s Hospital supplemental DMD newborn screening program in Boston with funding from CureDuchenne, have each contributed important data and expertise to this effort. Each pilot utilizes a slightly different approach to screening and following babies. This is important, as states often use different methods or different screening algorithms. Demonstrating that different screening algorithms work in different places provides evidence that newborn screening for Duchenne will work in diverse clinical settings.

The development of a RUSP nomination package is an intense effort that requires reviewing and compiling evidence on all therapies. Multiple workgroups composed of experts in Duchenne, newborn screening, and pediatrics have contributed to these efforts over the last year. PPMD is incredibly grateful to all the experts in Duchenne and NBS who have contributed to these efforts.

Founding President and CEO, Pat Furlong reflects on the importance of the organization's RUSP submission:

"This is a hopeful time in Duchenne therapy development. We have a robust experimental therapy pipeline targeting multiple physiological pathways. Yet therapeutic interventions are likely to be optimally effective the earlier they can be offered. Newborn screening is the most effective way to ensure that infants with Duchenne are diagnosed early, when therapies will likely be the most beneficial."

Ms. Armstrong goes on to explain how implementing NBS for Duchenne is critical to eliminating the long, expensive, and unnecessary diagnostic odyssey that most families experience:

"Despite many efforts, it still takes on average at least two years of uncertainty and concern for a family to get a diagnosis of Duchenne for their child. We know that there are families where the process has taken much, much longer and that where a person lives and where they get care also influence this. Individuals with Duchenne who are diagnosed at older ages have less benefit

from approved therapies and fewer opportunities to participate in research and clinical trials. Newborn screening is universal, which means it is equitable— it will ensure every baby is identified in time to consider treatments or participate in clinical trials."

PPMD is exceptionally grateful to the families, experts, and partners who have helped us to get to this point in our newborn screening efforts. Shares Ms. Furlong:

"Obviously no one wants their child to be diagnosed with Duchenne muscular dystrophy, but I am a firm believer that knowledge is power in our fight to end the progression of this fatal disorder. Recommendation to the RUSP will enable Duchenne newborn screening to be performed across the country, resulting in early diagnosis, early intervention, and most importantly, the best possible outcomes for these babies."