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Parent Project Muscular Dystrophy's Ground-Breaking Effort: Completion of a Successful Pilot in Newborn Screening for Duchenne Muscular Dystrophy

More than 36,000 Babies Screened in Partnership with New York State and NBSTRN; Data Presented at Association of Public Health Laboratories Newborn Screening Virtual Symposium

WASHINGTON, DC – October 6, 2021 – Parent Project Muscular Dystrophy (PPMD), a nonprofit organization leading the fight to end Duchenne muscular dystrophy (Duchenne), announced that the organization's Newborn Screening Pilot has successfully reached its completion, screening more than 36,000 babies born in New York State over the last two years. Data were presented today at the Association of Public Health Laboratories Newborn Screening Virtual Symposium about the pilot in New York State, which recently completed recruitment. As of July 31, 2021, 34 babies have been referred for significantly elevated levels of the muscle isoform of creatine kinase (CK-MM), suggestive of a muscular dystrophy. Four of these babies have been confirmed to have Duchenne/Becker muscular dystrophy, and one baby was identified as a carrier female.

This pilot testing program was launched in October 2019 in collaboration with a precompetitive consortium that includes the Newborn Screening Translational Research Network (NBSTRN) and New York State. The goal of PPMD's Newborn Screening Pilot was to prevent families from experiencing an unnecessary diagnostic odyssey and ensure that every family receives timely, supportive, accurate resources and clinical care at the time of diagnosis.

Families with babies identified to have Duchenne or any other muscular dystrophy through this pilot have access to expert genetic counseling and neuromuscular care, from the very beginning. They can benefit from early interventions and be prescribed appropriate therapies. They are eligible for clinical trials and research studies specific to infants with Duchenne. Families who participate in the pilot provide feedback on their experiences by completing surveys and an interview, in order to improve the newborn screening process for Duchenne. Moving forward, this pilot can serve as a model for newborn screening for all babies for Duchenne.

According to Founding President and CEO, Pat Furlong, PPMD has led a national effort to build a newborn screening infrastructure for Duchenne in the U.S. aimed at developing the evidence to support Duchenne newborn screening for the last seven years. Ms. Furlong says, "This is a hopeful time in Duchenne therapy development. We have a robust experimental therapy pipeline targeting multiple physiological pathways. Yet it is believed that therapeutic interventions may be optimally effective the earlier they can be offered. It is the goal of PPMD and the Duchenne community to help eliminate the diagnostic delay, the two or more painful, expensive and uncertain years that many families experience. Newborn screening is the most effective way to ensure that infants with Duchenne are diagnosed early, when therapies will likely be the most beneficial. We are grateful to all of our collaborators, including the NIH-funded Newborn Screening Translational Research Network (NBSTRN) and the New York State Department of Health, for all of their work on this pilot."

The pilot was funded and led by a consortium of Duchenne industry partners and PPMD with a commitment to early diagnosis and intervention in Duchenne. Current consortium members

include PTC Therapeutics, Sarepta Therapeutics, PerkinElmer, Solid Biosciences, Pfizer, Inc., and PPMD. The pilot is 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, and the Genetic Alliance's Expecting Health.

Niki Armstrong, PPMD's Newborn Screening Program Manager recognizes the monumental effort required from so many people and organizations to initiate this pilot and then continue it through in the midst of a pandemic. "This pilot has been a huge group effort, and we're incredibly grateful to all of those who have been willing to share their expertise, from the initial planning stages more than 5 years ago to the final analysis of the data, which is currently underway. And of course, we're deeply appreciative of our clinical sites at Northwell Health and New York Presbyterian Hospitals and the New York State Newborn Screening lab for their resourcefulness and resiliency, as they safely and successfully screened babies throughout a pandemic."

In addition to launching the Duchenne newborn screening pilot program, PPMD's newborn screening agenda includes active involvement on the reauthorization of the Newborn Screening Saves Lives Act; annual Duchenne-specific language within Appropriations and Report Language to ensure federal partners are focused on Duchenne newborn screening efforts; engagement with the federal Advisory Committee on Heritable Disorders for Newborns and Children; and leading the National Duchenne Newborn Screening Initiative, which has included the development of published care standards for newborns, ethical considerations for Duchenne newborn screening, and the publication of A Roadmap to Newborn Screening for Duchenne Muscular Dystrophy.

Ms. Furlong reflects on the completion of the pilot, "We are exceptionally grateful to the families, experts, and partners who have helped us to get to where we are today—four babies identified with Duchenne or Becker who now have access to early interventions and care. 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 deadly disorder. Without newborn screening, these families may have spent years on a stressful and exhausting diagnostic odyssey. Early diagnosis means early intervention, which will mean the best possible outcomes for these babies. This pilot, in combination with all of the past and ongoing pilots, lays the groundwork for Duchenne newborn screening to be performed across the country."

To learn more about PPMD's work in newborn screening, click here.

About Parent Project Muscular Dystrophy

Duchenne is a fatal genetic disorder that slowly robs people of their muscle strength. Parent Project Muscular Dystrophy (PPMD) is the largest, most comprehensive nonprofit organization in the United States focused on finding a cure for Duchenne—our mission is to end Duchenne.

We demand optimal care standards and strive to ensure every family has access to expert healthcare providers, cutting edge treatments, and a community of support. We invest deeply in treatments for this generation of Duchenne patients and in research that will benefit future generations. Our advocacy efforts have secured hundreds of millions of dollars in funding and won four FDA approvals.

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Everything we do—and everything we have done since our founding in 1994—helps those with Duchenne live longer, stronger lives. We will not rest until we end Duchenne for every single person affected by the disease. Join our fight against Duchenne at EndDuchenne.org and follow PPMD on Facebook, Twitter, and YouTube.

About the Newborn Screening Translational Research Network (NBSTRN)

The Newborn Screening Translational Research Network (NBSTRN) is funded by a contract from the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), National Institutes of Health (NIH) to the American College of Medical Genetics (ACMG) with the goal of advancing newborn screening related research (HHSN275201800005C). The NBSTRN is a key component of the Hunter Kelly Newborn Screening Research Program, and provides resources for investigators engaged in newborn screening-related research including new technology development, tools for developing the clinical history of genetic disorders and new treatment development. To learn more please visit www.nbstrn.org.

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