MDA testimony in front of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children on moving the Duchenne muscular dystrophy nomination to full evidence review – 2/10/23

Thank you for the opportunity to comment on today's deliberation on moving Duchenne muscular dystrophy forward to full evidence review. I am Paul Melmeyer, Vice President, Public Policy and Advocacy, at the Muscular Dystrophy Association. MDA is proud to serve the Duchenne, spinal muscular atrophy, and Pompe communities along with many other rare neuromuscular diseases.

Today we request the Committee vote to move the Duchenne muscular dystrophy nomination forward to full evidence review. MDA was proud to co-sponsor the nomination of Duchenne last summer, and under the leadership of Parent Project Muscular Dystrophy, provide the evidence to the Committee required for consideration.

We would like to emphasize several points as the Committee considers its vote.

First, we believe the evidence within, or referenced within, the nomination package is thorough and adequate to move the nomination forward. Duchenne is certainly a serious disease that would benefit from early diagnosis and early treatment. The progression of Duchenne is well understood due to decades of research funded by MDA, PPMD, and allied Duchenne organizations.

Second, MDA was pleased to co-fund a pilot study conducted in North Carolina by RTI International that tested the validity and reliability of using creatine kinase levels and follow up confirmatory genetic testing to screen for and diagnose Duchenne. This pilot study, along with studies in New York and Massachusetts, have shown the feasibility of screening for Duchenne at birth.

Third, there are several FDA-approved treatments available to individuals with Duchenne, including several exon-skipping therapies as well as corticosteroid treatments. We also anticipate a gene therapy to be approved by the FDA later this year for Duchenne. Like treatments in similar neuromuscular diseases, treating Duchenne early can help slow the progression of irreversible muscle loss and organ damage.

Finally a robust network of clinicians are prepared to offer comprehensive care to those who are newly diagnosed. Often these are the very same clinics treating infants newly diagnosed with SMA and Pompe, thus creating a familiarity within the neuromuscular disease clinical community for care and support of those diagnosed through newborn screening. These clinics are also usually familiar with any related neuromuscular disorder that might be caught through this screen.

In conclusion, we urge the Committee to vote to move Duchenne muscular dystrophy to full evidence review.