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 – 11/2/23

Thank you for the opportunity to comment on the ongoing review of Duchenne muscular dystrophy for consideration for the Recommended Uniform Screening Panel. 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 disease communities.

First and foremost, we are very grateful for the Committee's vote to move the Duchenne nomination on to Full Evidence Review in August's meeting. We hope the unanimous vote is at the very least evidence that our nomination deserves a full evaluation, and hope this is a key step in Duchenne soon joining the RUSP.

We are grateful for the time and effort expended by the Nominations and Prioritizations Workgroup as it thoroughly reviewed our nomination and came to the conclusion that Duchenne should move forward to full evidence review. We agreed with comments that pointed out that the current delay in clinical diagnosis hinders collection of treatment effectiveness data in earlier diagnosed individuals, but available evidence clearly points to earlier effectiveness, and the combination of ongoing clinical trials and implementation of newborn screening will fill any potential evidence gap.

As discussed in August, efforts to accelerate clinical diagnosis, and there have been many, have simply been unsuccessful even when symptoms present at an earlier timeframe. While improvements could be made around the edges, newborn screening in particular is uniquely capable of providing an earlier diagnosis.

We agree with comments that states should be well prepared to create their own validation processes and cutoffs for CK-MM testing just as states have done for many newborn screens currently universally included on state panels. We encourage this committee to empower and encourage states to create their own cut-offs and validating assays as this is something states and their programs are familiar with and capable to handle themselves.

Finally, we appreciate comments acknowledging the importance of new therapies coming to market, most notably Elevidys, a gene therapy, for boys with Duchenne ages 4 and 5. Furthermore, we strongly wish to re-emphasize comments noting the importance of an early diagnosis on life and family planning, PT and speech therapy options, and more.

Thank you again for the opportunity to comment today, and we look forward to Committee's ongoing review.