



**MDA testimony in front of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children on the Duchenne muscular dystrophy nomination – 5/4/23**

Thank you for the opportunity to provide comments and updates on our ongoing efforts to add Duchenne muscular dystrophy to 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 diseases.

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 were disappointed that the Committee voted to not move the Duchenne nomination to full evidence review in February, but we are undeterred in trying to move the nomination forward.

In addition to the points that Niki Armstrong with PPMD just updated you on pertaining to the availability of effective treatments for individuals with Duchenne, as well as the potential approval of a gene therapy for Duchenne later this month, and how important these treatments are for delaying the onset of many symptoms of Duchenne, we also wanted to provide updates and comments on several additional points raised by this Committee when discussing this nomination in February.

First, the Committee expressed concern about the availability of confirmatory testing for state newborn screening programs to confirm the diagnosis of Duchenne via next generation sequencing. Frankly, we do not share this concern as access to genetic confirmatory testing is not demonstrably different than the genetic testing to find the SMN2 gene for SMA, or confirm the genetic cause of Pompe disease. These genetic tests are substantially less expensive than they used to be and are fully accessible to state programs and providers. In addition to free genetic testing programs, genetic tests cost a

few hundred dollars at the most to find the genetic causes of Duchenne and related muscular dystrophies. With over 40 CLIA certified labs performing Duchenne genetic testing, and with this number expected to grow, this number is greater than labs conducting confirmatory testing for other RUSP-approved conditions.

We are also paying close attention to the evolving state policy environment pertaining to the use of dried blood spots. While several states are considering further limiting the use of dried blood spots in secondary research, law enforcement, or other venues, the use of dried blood spots for confirmatory testing within the initial newborn screening process is not something of concern as of yet.

Second, the Committee questioned the necessity of screening for Duchenne muscular dystrophy at birth, instead exploring the appropriateness of testing for Duchenne at a later date, perhaps the one-year wellness visit. We would strongly disagree with this approach. The presence of elevated CK levels in newborns with Duchenne is evidence that muscle damage caused by Duchenne is happening prior to birth and continues throughout the course of the disease. To intentionally delay a diagnosis only allows this muscle damage to continue unchecked for at least a year. Furthermore, according to CDC, anywhere from 10 to 30 percent of children don't have their well-child visits, with health system inequities exacerbating this further for minority populations.

Finally, without going further today during my testimony, we will also address questions pertaining to the false positive rate within the pilot studies, expectation of newborn screening for Duchenne at a population level, and more.

We look forward to addressing these and other concerns within our re-nomination of the package in the coming weeks, and are happy to answer any further questions. Thank you again for the opportunity to testify today.