September 12, 2025

CDR Leticia Manning, MPH,
Newborn Screening Team Lead,
Division of Services for Children with Special Health Needs,
Maternal and Child Health Bureau,
Health Resources and Services Administration
5600 Fishers Lane, Rockville, MD 20857

Re: Notice With Request for Comment: Consideration of Adding Duchenne Muscular Dystrophy to the Recommended Uniform Screening Panel

Dear Commander Manning,

On behalf of the Muscular Dystrophy Association (MDA) and Parent Project Muscular Dystrophy (PPMD), we submit this comment in strong support of adding Duchenne muscular dystrophy (Duchenne) to the Recommended Uniform Screening Panel (RUSP).

Duchenne is a devastating, progressive, and ultimately fatal genetic disorder that affects approximately 1 in every 5,000 live male births. Muscles are already being damaged by Duchenne at the time a child is born as evidenced by elevated creatine-kinase (CK)-MM in their blood. But without knowing the presence of Duchenne, most children are not clinically diagnosed until after the age of four, following years of irreversible muscle damage and loss.

Newborn screening is right for the Duchenne community: Currently, a diagnosis of Duchenne following a lengthy diagnostic journey results in missed opportunities for care, access to clinical trials, and treatment initiation. Current interventions largely focus on preserving healthy muscle, and the muscle loss caused by Duchenne likely makes interventions less effective than if they were administered earlier. By the time most boys are diagnosed, the optimal window for timely intervention has already closed.

Newborn screening offers the only path to achieving timely diagnosis for Duchenne. A variety of efforts over the preceding decades have attempted to decrease the age of diagnosis for Duchenne but none have succeeded. These include practice parameters from the American Academy of Neurology, a "National Task Force for Early Identification of Childhood NMD", and an American Academy of Pediatrics and Centers for Disease Control and Prevention toolkit. None of these efforts meaningfully altered the time to diagnosis for children with Duchenne.

As Duchenne is detectable and clinically present at birth, families deserve to know early and definitively so that they can access meaningful clinical interventions, FDA approved therapies, and coordinated multidisciplinary care proven to slow disease progression and improve quality of life. Physical and occupational therapy can be specifically tailored to children with Duchenne

as can guidelines for physical exercise. An earlier diagnosis can help mitigate the effects on families as Duchenne can cost hundreds of thousands of dollars in medical costs, an estimated 20 lost days of work a year for parents of children with Duchenne, and 17 out of state trips for medical care. Newborn screening also ensures that diagnosis is not dependent on where a child is born, family resources, or provider awareness.

Newborn screening meets all the parameters for Recommended Uniform Screening Panel inclusion: The evidence supporting the readiness and appropriateness of Duchenne for newborn screening is clear:

- **Feasibility:** Duchenne can be reliably detected through established and validated newborn screening methods. CK-MM testing, which is FDA-approved, is the standard approach for screening for the muscle damage caused by Duchenne. Tested in multiple pilot studies over several decades and now administered in Ohio and Minnesota's newborn screening programs, CK-MM as the newborn screen with genetic testing as the confirmatory test should minimize false positives and false negatives while successfully diagnosing infants with Duchenne. Over forty state newborn programs already have access to the equipment necessary to screen for Duchenne.

- **Clinical Actionability:** FDA-approved therapies are available, and earlier access improves outcomes. Corticosteroids and exon-skipping therapies are currently available to children at ages younger than the mean age of diagnosis, with other treatment modalities such as gene therapies likely to be available at younger ages soon. Physical and occupational therapy can be administered early as well as participation in clinical trials. Comprehensive standards of care are well established and can be initiated promptly following diagnosis.

- **Public Health Impact:** Early identification will transform the lives of affected children and families, while reducing the long-term health and economic burden on states and the healthcare system. Newborn screening has been shown to accelerate therapeutic development potentially leading to safer and even more effective therapies arriving at a more rapid pace.

Adding Duchenne to the RUSP reflects a commitment to early, timely diagnosis. It is the next critical step in ensuring that every child born with Duchenne has the best chance at a longer, healthier life.

We urge the Department of Health and Human Services and the Health Resources and Services Administration (HRSA) to recommend and approve the addition of Duchenne muscular dystrophy to the RUSP without delay.

Sincerely,

Sharon Hesterlee, PhD
interim President & CEO
Muscular Dystrophy Association

Pat Furlong
President
Parent Project Muscular Dystrophy