Consensus Statement in support of Duchenne Newborn Screening

We, as experts in the fields of genetics, pediatrics, neurology, and public health, hereby endorse the implementation of Duchenne muscular dystrophy (DMD) newborn screening as an essential component of comprehensive healthcare for infants. DMD is a severe and progressive genetic disorder that affects approximately 1 in 5,000 live male births, making it one of the most common and devastating childhood neuromuscular diseases.

After careful consideration of the available scientific evidence, we acknowledge the following key points supporting the implementation of Duchenne Newborn Screening:

Timely Intervention and Treatment Initiatives:

Duchenne is characterized by a progressive loss of muscle function, with symptoms typically appearing in early childhood. Timely diagnosis through newborn screening provides a critical window for intervention and initiation of therapeutic strategies before irreversible muscle damage occurs.

Timely treatment, including early intervention services that address delays in speech, motor, and cognitive development, as well as emerging genetic and pharmacological therapies, have shown promise in slowing disease progression and improving outcomes for individuals with Duchenne. Newborn screening facilitates timely access to these interventions, maximizing their potential benefits.

Additionally, families of babies identified with Duchenne through newborn screening benefit from genetic counseling resources, as well as opportunities for emotional and psychological support, allowing for informed decision-making and proactive healthcare planning.

Reduced Diagnostic Delays and Avoidable Diagnostic Costs:

Newborn screening eliminates the diagnostic odyssey that many families currently face, reducing the time from symptom onset to diagnosis. While medical costs are unavoidable in Duchenne, the avoidable costs attributable to delayed diagnosis represent the burden on patients and families searching for a diagnosis. Delayed diagnosis also results in healthcare dollars that could be better spent on treatment and supportive therapies that improve patient quality of life and may even increase workforce productivity for families. Recently, a study of the avoidable costs in the Duchenne diagnostic odyssey were $211, 229 per family and averaged 20 days of missed work per year for a parent.

Public Health Impact:

Implementing Duchenne Newborn Screening contributes to the broader public health goal of early detection and intervention for genetic conditions. It aligns with the principles of preventive medicine and represents a proactive approach to managing a significant healthcare challenge.

Ethical Considerations:

> The benefits of early diagnosis and intervention, coupled with the minimal invasiveness of newborn screening, support the ethical justification for implementing Duchenne Newborn Screening. Informed consent and appropriate counseling must be integral components of the screening process, ensuring respect for autonomy and shared decision-making.

Based on the accumulating evidence, we strongly advocate for the incorporation of DMD into newborn screening programs. Timely identification of affected individuals is a crucial step towards eliminating unnecessary and avoidable diagnostic odyssey, improving patient outcomes, and enhancing the quality of life for children with Duchenne. We call upon policymakers, healthcare providers, and advocacy groups to work collaboratively in implementing and optimizing Duchenne Newborn Screening programs to ensure their effectiveness and accessibility on a national scale.

Signed,

Abigail N. Schwaede, MD
Attending Physician, Neurology, Lurie Children's Hospital of Chicago
Assistant Professor of Pediatrics (Neurology and Epilepsy), Northwestern University Feinberg School of Medicine

Alex Fay MD,PhD
Associate Professor of Neurology
Child Neurology and Neuromuscular Medicine
UCSF Benioff Children's Hospital

Alexander Zygmunt, MD
Assistant Professor, Child Neurology, Comprehensive Neuromuscular Center
Cincinnati Children's Hospital Medical Center/University of Cincinnati Medical Center

Amy Harper, MD
Professor, Pediatric Neurology, Department of Neurology
Children's Hospital of Richmond at VCU

Anne M. Connolly, MD FAAN
Robert F. and Edgar T Wolfe Chair
Professor of Pediatrics and Neurology
Chief of Pediatric Neurology
Nationwide Children's Hospital
The Ohio State University College of Medicine

Aravindhan Veerapandiyan, MD
Associate Professor of Pediatrics
University of Arkansas for Medical Sciences College of Medicine
Director, Comprehensive Neuromuscular Program
Director, PPMD Certified Duchenne Care Center
Co-Director, MDA Pediatric Care Center
Arkansas Children's Hospital

Arlene Deseo, RN, BSN
Multidisciplinary Neuromuscular Program
Nurse Care Manager
Children's Hospital Los Angeles

Ashlee Bolger, MD, MEd
Associate Professor- Clinical Pediatrics and Physical Medicine and Rehabilitation
Cincinnati Children's Hospital/University of Cincinnati

Ashley Stanley-Copeland, MD
Assistant Professor, Department of Neurology
Pediatric Neurology / Neuromuscular Medicine
Dell Medical School, The University of Texas at Austin

Barry Byrne, MD
University of Florida

Basil T. Darras, MD
Director, Neuromuscular Program
Boston Children's Hospital

Bo Lee, MD
University of Rochester

Brenda Wong, M.D.
Professor of Neurology and Pediatrics
Director, DMD Program
U Mass Chan Med School

Carolina Tesi Rocha, MD
Clinical Professor of Neurology & Neurological Sciences, Stanford University
Division of Child Neurology and Neuromuscular Medicine
Director, Pediatric Neuromuscular Clinic

Chamindra G. Laverty, M.D.
Clinical Professor of Neurosciences
University of California, San Diego

Craig M. McDonald, MD
Professor and Chair
Department of Physical Medicine & Rehabilitation
Professor of Pediatrics
Director MDA Neuromuscular Disease Clinics
University of California Davis Health

Craig Zaidman, MD
Professory of Neurology
Department of Neurology Divisions of Child Neurology and Neuromuscle Washington University in St. Louis School of Medicine

Cristian Ionita, MD
Associate Professor of Clinical Pediatrics (and Neurology)
Department of Pediatrics
Yale University, School of Medicine

Crystal Proud, MD
Children's Hospital of the Kinds Daughter

Cuixia Tian, MD
Associate Professor of Pediatrics and Neurology
Co-director of Neuromuscular Comprehensive Care Clinic
Director, MDA clinic
Director, Certified Duchenne Care Center
Division of Neurology
Cincinnati Children's Hospital Medical Center

Dana O'Rourke, MD
Attending Neurologist
Children's Hospital Los Angeles
Assistant Professor of Neurology and Pediatrics
Keck School of Medicine
University of Southern California

Daniel Guillen, MD
Pediatric Neuromuscular Neurologist

David Dredge, MD
Director, Pediatric Neuromuscular Clinic
Mass General Hospital

Diana Castro, MD
Board Certified Neurologist and Nueromuscular Physician
Founder and Director
Neureology and Neuromuscular Care Center/Neurology Rare Disease Center

Diana L. Marchese, MD
Pediatric Rehabilitation Medicine
Assistant Professor of Pediatrics
Children's Mercy Kansas City
UMKC School of Medicine

Edward C. Smith, MD
Medical Director, RDR-NC
Principal Investigator
Rare Disease Research

Elizabeth McNally MD PhD
Elizabeth Ward Professor and Director
Center for Genetic Medicine
Northwestern University
Feinberg School of Medicine

Emma Ciafaloni, MD, FAAN
Robert C. and Rosalyne H. Griggs Professor in Experimental Therapeutics of Neurologic Disease
Professor of Neurology, Pediatrics and Obstetrics and Gynecology
Associate Chair of Research
Director Pediatric Neuromuscular Medicine

Garey Noritz, MD, FAAP, FACP
Division Chief, Complex Health Care Program
Professor, The Ohio State University
Nationwide Children's Hospital Columbus, OH
Han C. Phan, MD
Director of Clinical Research
Rare Disease Research

Hoda Z. Abdel-Hamid, MD, MSc, FAAN
Associate Professor of Pediatrics, Neurology and Clinical and Translational Science
Division of Child Neurology
University of Pittsburgh
Director, Pediatric Neuromuscular Program
Director, Pediatric EMG Laboratory
Director, Pediatric MDA Clinic, PPMD CDCC and CMT centers of excellence

Jaclyn Omura, MD
Pediatric Rehabilitation Medicine Physician
Co-Director, Neuromuscular Program
Seattle Children's Hospital

Jonathan B. Strober, M.D.
Director, Pediatric Neuromuscular Clinic and MDA Care Center Professor, Clinical Neurology & Pediatrics
UCSF Benioff Children's Hospital San Francisco

Kaitlin Batley, MD
Director, Pediatric Neuromuscular Program
Assistant Professor of Pediatrics and Neurology
UT Southwestern Medical Center and Children's Health

Katherine D. Mathews, MD, FAAN
Vice Chair for Clinical Investigation, Department of Pediatrics
Professor, Departments of Pediatrics and Neurology
University of Iowa Carver College of Medicine

Leigh Maria Ramos-Platt, MD
Director Multidisciplinary Neuromuscular Program
Children's Hospital Los Angeles

Loren Santiago, MSN, NP
Multidisciplinary Neuromuscular Program
Nurse Care Manager
Children's Hospital Los Angeles

Mathula Thangarajh, MD, PhD
Virginia Commonwealth University

Matt Elrick, MD, PhD
Assistant Professor
Center for Genetic Muscle Disorders, Kennedy Krieger Institute
Pediatric Neurology & Neuromuscular Medicine, Johns Hopkins School of Medicine

Megan Waldrop, MD
Assistant Professor of Pediatrics and Neurology, Ohio State University
Principal Investigator, Center for Gene Therapy, Neuromuscular Division
Nationwide Children's Hospital

M. Carrie Miceli, Ph.D.
Professor of Microbiology, Immunology and Molecular Genetics
Co-Director, Center for Duchenne Muscular Dystrophy
David Geffen School of Medicine and College of Letters and Sciences at UCLA

Migvis Monduy, MD
Pediatric Neurologist
Director Neuromuscular Program
Nicklaus Children's Hospital

Monika Krzesniak-Swinarska, MD
Associate Professor of PM&R, Neuromuscular, Neurology
Adult MDA Care Center Director
EMG Lab Director
Department of Neurology
Department of Orthopedics & Rehabilitation
University of New Mexico Health Sciences Center

Nancy Kuntz, MD, FAAN
Attending Neurologist, Ann & Robert H. Lurie Children's Hospital of Chicago
Medical Director, Mazza Foundation Neuromuscular Disorders Program,
PPMD-certified Duchenne Care Center and MDA Care Center
MDA Clinic Director, MDA Clinic at Ann & Robert H. Lurie Children's Hospital of Chicago
Professor of Pediatrics and Neurology, Northwestern University Feinberg School of Medicine

Noelle Tiongson, MD
Attending Neurologist
Children's Hospital Los Angeles
Assistant Professor of Neurology and Pediatrics
Keck School of Medicine
University of Southern California

Omer Abdul Hamid, MD
Nemours Children's Health

Partha S. Ghosh, MD
Associate Professor of Neurology
Harvard Medical School
Director, EMG Laboratory
Boston Children's Hospital

Paul R. Carney, MD
Director, Division of Pediatric Neurology
Professor of Pediatrics and Neurology
Professor of Biomedical Engineering by Courtesy
University of Missouri School of Medicine

Perry Shieh, M.D. Ph.D.
Professor of Neurology and Pediatrics
David Geffen School of Medicine at UCLA

Peter B. Kang, MD
Professor and Vice Chair of Research, Department of Neurology
Director, Greg Marzolf Jr. Muscular Dystrophy Center
Scholar, Institute for Translational Neuroscience (ITN)
University of Minnesota

Peter Karachunski, MD, Clinical Director
Greg Marzolf Jr. Muscular Dystrophy Center
University of Minnesota and MDA Care center
Division of Pediatric neurology
Department of Neurology

Randal Richardson, MD, MMS
Neuromuscular Neurologist/MDA Clinic Director
Gillette Children's Specialty Healthcare

Raymund David, MD
Neuromuscular director
Valley Children's Hospital

Renata Shih, MD
Associate Professor of Pediatrics
Child Health Research Institute
University of Florida

Robert Lopez Alberola, M.D.
Associate Professor of Neurology and Pediatrics
Chief, Division of Child Neurology
Director of Pediatric MDA Clinic
University of Miami School of Medicine

Robert C. Griggs, MD FACP, FAAN (Honorary)
Professor of Neurology, Medicine, Pathology and Laboratory Medicine, and Pediatrics,
University of Rochester

Russell Butterfield MD, PhD
Associate Professor of Child Neurology
Director Neuromuscular Clinics,
University of Utah, School of Medicine

Sabrina Yum, M.D.
Associate Professor of Pediatrics and Clinical Neurology
Perelman school of Medicine at the University of Pennsylvania
Attending, Pediatric neurology/neuromuscular medicine
Program Director, Neuromuscular Fellowship program
Children's Hospital of Philadelphia

Samuel Mackenzie, MD, PhD
Assistant Professor
Neuromuscular Disease and Child Neurology
University of Rochester Medical Center

Saunder M. Bernes M.D.
Associate Clinical Professor Child Health
University of Arizona College of Medicine

Stanley F. Nelson, MD
Dr. Allen and Charlotte Ginsburg Endowed Chair in Translational Genomics
Distinguished Professor of Human Genetic, Pediatrics, Neurology, Psychiatry, and Pathology and Laboratory Medicine
Director, California Center for Rare Diseases
Co-Director, Center for Duchenne Muscular Dystrophy
David Geffen School of Medicine at UCLA

Stephen Chrzanowski MD, PhD
Neuromuscular Fellow
Department of Neurology, Neuromuscular Division, Boston Children's Hospital, Brigham and Women's Hospital, and Massachusetts General Hospital

Stephanie Manberg, DO
Pediatric Neuromuscular Specialist
University of Utah
Susan Apkon, MD
Professor and Vice-Chair, Department of Physical Medicine and Rehabilitation
University of Colorado School of Medicine

Urvi Desai
Director, Carolinas MDA Care Center (Adult and Pediatric)
Atrium Health Wake Forest University

W. Bryan Burnette, MD, MS
Professor of Clinical Pediatrics and Neurology
Vanderbilt University Medical Center

Wendy K. Chung, MD PhD
Chief, Department of Pediatrics
Boston Children's Hospital
Harvard Medical School

Vamshi K. Rao, MBBS, MD
Associate Professor of Pediatrics

Yaacov Anziska MD
Associate Professor of Neurology
SUNY-Downstate Medical Cente