National Foundation for Women Legislators

2024 NFWL Annual Conference Resolution

Introduced November 2024

Resolution calling upon states to add Duchenne muscular dystrophy to their respective newborn screening panels without delay to ensure health equity, improve the quality of life for patients, and save lives with a simple bloodspot.

WHEREAS, Duchenne muscular dystrophy (Duchenne) is a rare life-limiting pediatric neuromuscular genetic disease that affects about one in 5,000 newborn males (about 20,000 new cases each year worldwide);

WHEREAS, since the Duchenne gene is found on the X-chromosome, it primarily affects boys, however, females can be carriers for Duchenne and can manifest varying ranges of physical symptoms of Duchenne;

WHEREAS, Duchenne is caused by a mutation in the gene that encodes for dystrophin;

WHEREAS, without dystrophin, muscles deteriorate and are not able to function or repair themselves properly, and patients irreversibly lose muscle starting in utero;

WHEREAS, children living with Duchenne experience progressive muscle weakness that worsens over time and typically leads to a loss of the ability to walk in their early teens, use their arms in their teens, and then breathe unassisted in their twenties;

WHEREAS, young men with this debilitating disease typically only live into their mid- to late twenties;

WHEREAS, with the ability to affect anyone, Duchenne muscular dystrophy can be passed from parent to child, but approximately 35% of cases occur because of a random spontaneous genetic variation;

WHEREAS, there is consensus that the pace of progress in newborn screening lags when compared to advances in treatments for genetic conditions and Duchenne is no exception;

WHEREAS, the average age of diagnosis has remained five years old for the last thirty years despite focused efforts to achieve earlier diagnosis by the Centers for Disease Control and Prevention and others;

WHEREAS, there are now eight U.S. Food and Drug Administration (FDA) approved treatments for Duchenne, seven of which are indicated for patients below the average age of diagnosis, including a gene therapy, and an FDA-authorized newborn screening test for Duchenne;

WHEREAS, although a process exists for the review and addition of conditions to the federal Recommended Uniform Screening Panel (RUSP) used by the United States Department of Health and

Human Services, that process currently takes an average of six years from original nomination to add conditions to the RUSP;

WHEREAS, due to unnecessary delays in the federal process and the irreparable damage that results from a delayed Duchenne diagnosis, five states—Ohio, New York, Minnesota, Illinois, and Massachusetts—have proactively added Duchenne to their respective state newborn screening panels;

WHEREAS, in the absence of treatment, Duchenne robs the body of muscle every day and that muscle is lost forever;

WHEREAS, delayed diagnosis of Duchenne has a profound impact on patients and their families, who report noticing symptoms more than 2 years before receiving an official diagnosis;

WHEREAS, patients from minority populations experience a significantly longer delay in diagnosis, exacerbating health disparities;

WHEREAS, the EveryLife Foundation's study "The Cost of Delayed Diagnosis in Rare Disease"[1] found that members of the Duchenne patient community suffered an economic impact of over $200,000 in total medical costs, seeking specialists, and loss of productivity per family and the average caregiver loses 20 days of work each year while seeking out a diagnosis for Duchenne;

WHEREAS, Duchenne newborn screening ensures timely diagnosis, supports health equity, empowers families to make informed decisions about necessary care, provides the opportunity for early treatment, improves health outcomes for patients, and reduces long-term health care costs;

THEREFORE, BE IT RESOLVED, that the National Foundation of Women Legislators (NFWL) recognizes the need for newborn screening for Duchenne muscular dystrophy to ensure timely diagnosis for this devastating, rare pediatric neuromuscular disease;

BE IT FURTHER RESOLVED, that the NFWL acknowledges the availability of transformative treatments for Duchenne and that for patients living with Duchenne, time is muscle; and

BE IT FINALLY RESOLVED, that the NFWL calls upon states to add Duchenne to their respective newborn screening panels without delay to ensure health equity, improve the quality of life for patients, and save lives with a simple bloodspot.

[1] EveryLife Foundation, *The Cost of Delayed Diagnosis in Rare Disease: A Health Economic Study* (Sept. 2023), https://everylifefoundation.org/wp-content/uploads/2023/09/EveryLife-Cost-of-Delayed-Diagnosis-in-Rare-Disease_Final-Full-Study-Report_0914223.pdf.