Public Health Committee
JOINT FAVORABLE REPORT

Bill No.: HB-5321
Title: AN ACT REQUIRING NEWBORN SCREENING FOR DUCHENNE MUSCULAR DYSTROPHY.
Vote Date: 3/11/2024
Vote Action: Joint Favorable Substitute
PH Date: 3/6/2024
File No.: 86

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SPONSORS OF BILL:

The Public Health Committee

REASONS FOR BILL:

Duchenne Muscular Dystrophy (DMD) is one of the most severe and debilitating forms of this hereditary condition. DMD causes progressive muscle degeneration and weakness, and is usually seen between ages 2 and 3, primarily affecting males. This bill would require all newborns to be tested for this disorder as part of the newborn screening process approved by the Department of Public Health (DPH). Currently, newborn screenings are performed 24 to 48 hours after birth in health care institutions, by licensed nurse- midwives, and by midwives using blood spot specimens. This bill would add DMD to the list of conditions tested. Substitute language in the original bill moved the beginning date for this testing from July 1, 2026, to July 1,2027.

RESPONSE FROM ADMINISTRATION/AGENCY:

Manisha Juthani, Commissioner of the Department of Public Health (DPH):
Dr. Juthani shared that the federal government is currently reviewing the addition of DMD to the Recommended Uniform Screening Panel (RUSP) for newborns. This addition has strong support. In Connecticut, our newborn screening program includes any disorder listed on the RUSP. If this bill were to pass in its current form, it would result in a significant cost to the state since this funding is not part of the Governor's proposed budget. However, if we wait for the federal government to add DMD to the RUSP, then DPH will be able to use funds from the federal Health and Resources Propel Grant (HRSA) to aid in the cost of implementing the additional screening. The Department appreciates and supports the intent of the bill but cannot support it at this time.

Mitch Bolinsky, State Representative, 106th District, CT General Assembly:
Representative Bolinsky supports this bill.

NATURE AND SOURCES OF SUPPORT:

Kelly Maynard, President and Co-Founder, Little Hercules Foundation:
Ms. Maynard testified on behalf of advocacy organizations whose purpose is to improve the lives of those diagnosed with DMD. These organizations provide education, awareness, family assistance as well as support for medical care and research to help families of children with DMD. This rare and progressive disorder afflicts newborns at a rate of one in 3,500 births and despite efforts to enhance early detection, the average age remains around five years. Currently, there are seven FDA-approved treatments for DMD. One of which is a groundbreaking gene therapy with five more promising gene therapies in development. Screening and early detection is extremely important to allow effective treatments to begin as soon as possible. Delayed diagnosis results in healthcare dollars that could have been better spent on treatment and supportive therapies.

Dr. Gyula Acsadi, Chief of Pediatric Neurology, Connecticut Children's Medical Center:
Dr. Acsadi pointed out that diagnosis of DMD is often delayed and not identified in patients until between the ages of 5 -7years. By this age, irreversible muscle damage has already occurred. If earlier diagnosis had been established, it would have allowed for more timely treatment and management of symptoms. Dr. Acsadi shared that the center is currently working with several families each with two affected DMD boys. The families were unaware of the disease in the first child before they had the second child. Early detection would have allowed for timely treatment and counseling for family planning.

Debra Riley, Elementary School Special Education Teacher, and Board-Certified Behavior Analyst:
As a special education teacher, Ms. Riley has worked with students who have intense special needs including children diagnosed with DMD at birth, as well as students who were not diagnosed by the age of 5 or 6 and entering kindergarten. The earlier a family could be provided with a diagnosis the better the outcome. Many families endure a long, costly, and painful road to diagnosis which sometimes includes counterproductive interventions. This screening would provide a cost-effective transformative tool positively impacting the lives of the children and empowering their families to cope with this debilitating condition.

Lindsey DeVito, Mother:
Ms. DeVito has two hard-of-hearing children who were diagnosed at birth. Her children, now ages 10 and 13, are thriving due to the CT Early Hearing Detection and Intervention Program which is part of the newborn screening panel performed on children just after birth. If this screening had not been conducted, her children could have gone through their most developmentally formative years undetected. Children born with undiagnosed DMD might go through these very important early years losing critical time during which families could have sought interventions and supports to give their children the chance for a better future.

The following individuals provided testimony in which they shared their personal experiences having children with DMD and the overwhelming impact it has had on the quality of life for their families. All strongly support this bill. The screening of newborns shortly after birth is a proactive tool that would have had a profoundly positive impact on these families facing this debilitating condition:

- Pat Furlong, Founding President, and CEO of Parent Project Muscular Dystrophy.
- Kathryn White, Mother of 6-year-old Theodore with DMD.
- Audra Carbone, Mother of 10-year -old Kevin living with DMD.
- Liz Duffy, Mother of 6.5-year-old Ned who was diagnosed with DMD at age 2 years.
- Rasha Alnaibari, Mother of a boy who began to show symptoms of DMD at six months but was not properly diagnosed until years later losing valuable time to treat this condition.

NATURE AND SOURCES OF OPPOSITION:

None expressed.

Reported by: Katheen Panazza **Date: March 12, 2024**